Richard Cohen
Alastair Windsor
Editors

Anus

Surgical Treatment and Pathology

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For our wives Madeleine Cohen and Lily Windsor

Foreword

The reader might wonder why another textbook on the anus is needed. There are three main reasons. First anal disease is exceedingly common in all parts of the world. Secondly treatment is effective in most cases, but when surgery is performed badly the patient can be disastrously and often irreversibly worse. Thirdly new developments in the understanding and treatment make proctology a dynamic field for research and its application to management.

The book is not on colon and rectal surgery but rather on proctology in which anal pathology is set within the context of wider disease where relevant. "*Anus*" is edited by two internationally known and experienced surgeons who have themselves contributed to knowledge in the field through research, publications and teaching. They have brought together authors from all over the world, who are recognized practitioners in the field. The reader is given an accurate account of the normal structure and function of the anus including clinical and radiological anatomy and physiology. The importance of histopathology is dealt with in a chapter dedicated to diagnosis and the subsequent chapters then follow from congenital anomalies to neoplasia, prolapse, incontinence, haemorrhoids, fissure and anorectal sepsis. There is a section on dermatological conditions and sexually transmitted disease with the book ending with an account of plastic procedures around the anus.

Every attempt has been made to incorporate new information regarding etiology, diagnosis and treatment to give the reader an up-to-date account of the subject. The use of Tables and Boxes to present data makes the book easy to read. Many of the chapters contain high quality photographs to illustrate diseases particularly those due to dermatological and sexual transmission. The chapters on radiological pathological anatomy contain excellent illustrations which are very useful since they summarise the various pathological forms of anorectal sepsis and prolapse, enabling the reader to learn the radiological counterpart of clinical examination . Each chapter contains a useful bibliography which will in itself be a valuable resource enabling the reader to gain access to the literature including classic publications and recent developments.

The book will be valuable for consultant practitioners in proctology, but it will also help colorectal surgeons, gastroenterologists, trainees, specialist nurses and students who are interested in increasing their knowledge. Proctology is in many ways still a largely clinical speciality and the theme throughout the

book implicitly recognizes this while at the same time it demonstrates where investigations are either essential or likely to help clinical decision taking. It is an authoritative, informed and up-to-date contribution to an important area of disease.

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The Normal Anus

1

Alexis M.P. Schizas and Andrew B. Williams

Anal Canal Anatomy and Histology

The anal canal begins at the anorectal junction as the rectum passes through the pelvic cavity at the cranial end of the levator ani muscle forming a sharp angle. The anal canal in adults is between 2.5 and 5 cm in length extending caudally to terminate at the anal verge [1].

The anal canal is derived from the caudal hindgut which is divided by the urorectal (cloacal) septum into the urogenital sinus (giving rise to the urethra and bladder) and rectum [2]. The rectum and superior anal canal are separated from the outside by the anal membrane which breaks down by the end of the eighth week of gestation. Abnormal separation of the cloaca, with the anal canal posteriorly and the urethra and bladder anteriorly, results in a variety of anorectal malformations. An abnormality in the development of the urorectal septum in a posterior direction leads to the majority of the anorectal abnormalities such as rectal atresia and fistulas between the rectum and urethra, urinary bladder or vagina.

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The anal canal epithelium provides the transition point between the proximal rectal intestinal columnar epithelium to the distal stratified epithelium of the skin.

The rectum has a thick muscularis propria layer, containing an inner circular and outer longitudinal muscle layer of rectal muscle, which is capable of peristaltic activity. The muscularis mucosa is also a prominent feature of the rectum producing a rhythmic contraction which prevents clogging of the glands and enhances expulsion of mucus. The rectal epithelium has numerous mucus-secreting goblet cells that lubricate the stool and absorptive cells to absorb water from the stool which are arranged in closely packed tubular glands, greatly increasing surface area.

The anal canal epithelium is divided in to three sections. The proximal canal (cranial end of puborectalis to the cranial end of the dentate line) is lined by simple columnar epithelium. The epithelium in the proximal section is similar to that of the rectum but has shorter more irregular crypts and more smooth muscle fibres in the lamina propria layer. This changes to stratified squamous epithelium lower in the anal canal along an intermediate transition zone just above the dentate line [3]. The transition zone epithelium (zone between uninterrupted columnar mucosa above and uninterrupted squamous epithelium below) has a variable length of between 3 and 11 mm. In the fetus and newborn, there is a sharp transition, but in the adult this transition is not so sharp and stratified squamous epithelium may be present on columns. The transitional zone epithelium

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produces minimal mucin and may have features of squamous epithelium but contains anal glands and endocrine cells in the submucosal layer. The distal zone (caudal end of the dentate line to the anal verge) contains non-keratinizing squamous epithelium. The epithelium at this level does not contain glands or hair follicles. The squamous epithelium of the anal canal merges with perianal skin (with keratin, hair follicles and apocrine glands) at anal verge.

The dentate (pectinate) line is at the transitional zone and identified at the caudal end of the anal valves where they form to create a circumferential line. The anal mucosa forms between five and ten anal columns (columns of Morgagni) cranial to the dentate line. The columns are separated by anal valves which are easily identifiable in children but are more difficult to identify in adults [2]. The anal valves contain the rectal venous plexus. The anal glands discharge via the anal crypts at the level of the dentate line. The anal crypts are not present below the dentate line.

The layer deep to the anal mucosa is formed by subepithelial tissue, which is composed of connective tissue and smooth muscle [1]. Anal (vascular) cushions are a normal structure of anal canal, found within this subepithelial layer; these contribute to anal closure. The cushions contain blood vessels, connective tissue and smooth muscle. The anal cushions are thought to be important in the maintenance of continence and increase in thickness throughout life.

Beneath the subepithelial layer, the caudal continuation of the circular smooth muscle of the rectum thickens to form the internal anal sphincter [3]. The internal anal sphincter begins as the circular smooth muscle passes through the cranial end of puborectalis and terminates caudally in a clearly defined edge at a variable distance from the anal verge.

The continuation of the longitudinal muscle layer of the rectum forms the longitudinal muscle layer in the anal canal and lies between the internal and external anal sphincters in the intersphincteric space. The anal glands that open via the anal crypts along the dentate line are found in the intersphincteric space. The longitudinal muscle comprises smooth muscle cells from the outer layer of the rectal wall and striated muscle from levator ani, puborectalis and pubococcygeus. Caudally the fibres from this layer pass through 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, giving the radial corrugation of perianal skin.

The external anal sphincter is made up of striated muscle and surrounds the longitudinal muscle forming the outer border of the intersphincteric space. Figure 1.1 shows a sagittal view of the anal canal. The widely held opinion is that the external anal sphincter is a tripartite structure [4]. In this arrangement the external sphincter is divided into deep, superficial and subcutaneous parts. The deep section of the external anal sphincter is fused with puborectalis, with the deep and subcutaneous parts of the sphincter forming rings of muscle, between them elliptical fibres from the superficial part of the external anal sphincter run anteriorly from the perineal body to the coccyx posteriorly. Other models of the external anal sphincter describe it as a single muscle continuous with the puborectalis muscle [3], or one that has a two-part structure [5]. The two-part model suggested a deep external anal sphincter and a superficial external anal sphincter. The two-part model corresponds to the puborectalis and deep external anal sphincter combined and the fused superficial and subcutaneous sphincter of the tripartite model. The external anal sphincter muscle may not be complete in certain places, and it has been suggested that the smooth and skeletal muscles form an intimately integrated anal sphincter complex.

Imaging of the anal canal using anal endosonography (AES) and magnetic resonance imaging (MRI) has not completely resolved the anatomical question regarding the external anal sphincter. Anal canal imaging in women suggested that the anterior external anal sphincter is shorter than the posterior and that the lateral external anal sphincter length was longer than the anterior/posterior length, whereas a more cylindrical uniform shape was present in males. It is however more likely that the women simply have a shorter external anal sphincter than males and it does not actually differ in configuration [6].

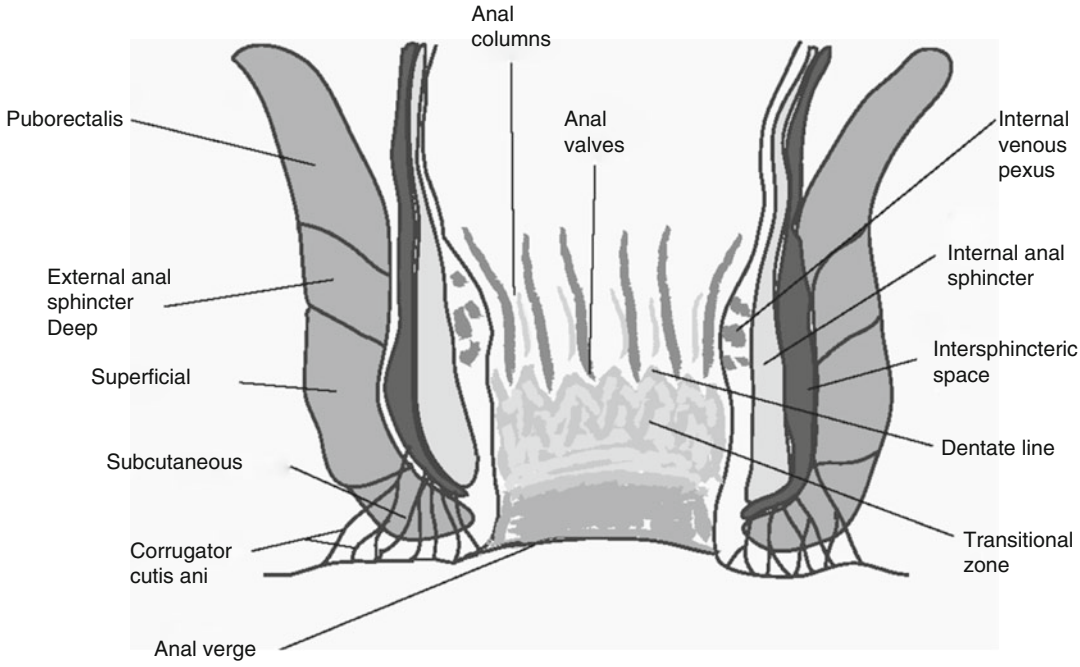

Fig. 1.1 Sagittal view through the normal anal canal

The external anal sphincter is innervated by the pudendal nerve (S2–S4) [1], which leaves the pelvis via the lower part of the greater sciatic notch, where it passes under the lower border of the piriformis muscle. It then crosses the ischial spine and sacrospinous ligament to enter the ischioanal 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 (mainly S2 and S3), 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. In monkeys there is a degree of cross-innervation between the two sides of the sphincter, although the degree of overlap in humans is limited. 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 fibres to the superior and inferior hypogastric plexuses, and the parasympathetic supply is from the second to fourth sacral nerve roots via the nervi erigentes [1]. Fibres 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.

The blood supply to the anal canal cranially, in keeping with its hind gut origin, is from the superior rectal artery (branch from the inferior mesenteric artery) as its terminal branches pass from the rectum to the anal canal between the mucosa and muscle layers. These arteries anastomose with the middle rectal artery, a branch of the internal iliac artery. The inferior rectal artery is a branch from the internal pudendal artery arising from the lateral wall of the ischioanal fossa, crossing the fossa to supply the caudal anal canal muscles and skin. The inferior rectal artery anastomoses with the middle and superior rectal arteries in the anal canal.

Venous drainage of the anal canal is via the internal venous plexus in the submucosa and

external plexus outside the muscular layer. The internal plexus drains mainly to the superior rectal veins and then into the inferior mesenteric veins. The external and to a lesser extent the internal plexuses drain into the inferior rectal vein which drains into the internal pudendal vein. The internal venous plexus is a site of a portocaval venous anastomosis which may lead to varices in portal hypertension.

The lymphatic drainage of the cranial anal canal is via the anorectal then perirectal lymph nodes to the preaortic lymph nodes. The lymphatic drainage to caudal anal canal is to the internal and common iliac lymph nodes. The lymphatic drainage of the skin of the anal canal is to the medial superficial inguinal lymph nodes.

Anal Canal Physiology

Anal 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 defaecation 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 defaecation.

As a result of the complex interplay between the factors involved in continence and faecal evacuation, a wide range of investigations are 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.

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. Between 60 and 85 % of resting anal pressure is attributed to the action of the internal anal sphincter [7]. The external anal sphincter and the puborectalis muscle generate maximal squeeze pressure [8]. 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 faeces are due to external sphincter problems.

The function of the internal anal sphincter is under autonomic control. The sympathetic nerve supply to the internal anal sphincter is excitatory, and the parasympathetic supply is inhibitory. Sympathetic nervous activity is thought to enhance and parasympathetic activity to reduce internal sphincter activity. Relaxation of the internal anal sphincter may be mediated via non-adrenergic, non-cholinergic nerve activity via the neural transmitter nitric oxide. The internal anal sphincter is in a state of tonic contraction; as the rectum distends, to a certain volume, there is reflex relaxation of the internal anal sphincter and followed by a contraction of the external anal sphincter (rectoanal inhibitory reflex).

The rectum, most of the time, is empty as a result of the angulation at the rectosigmoid junction. As the volume of faeces increases in the sigmoid colon, peristalsis forces faeces into the rectum. The volume required to elicit the rectoanal inhibitory reflex correlates with the volume required for first sensation [9]. As the rectal wall distends, afferent signals spread through the myenteric plexus and initiate a peristaltic wave in the descending colon, sigmoid colon and rectum. The internal anal sphincter then relaxes, due to inhibitory signals from the myenteric plexus, and contents from the rectum enter the anal canal and to come in contact with the transition zone mucosa, allowing discrimination of solid from fluid and flatus, a process vital to maintaining continence. In order to prevent incontinence there is a reflex contraction of the external anal sphincter during this anorectal sampling process. The rate of recovery of sphincter tone after this relaxation differs for the proximal and distal canal, which may be important in maintaining continence [10]. In patients with Hirschsprung's disease, progressive

systemic sclerosis, Chagas disease and initially after a coloanal anastomosis, there is no reflex relaxation of the internal anal sphincter. Further studies investigating the role of the rectoanal inhibitory reflex have shown that there is a greater sphincter relaxation of the anal sphincters in incontinent patients as rectal volume increases and patients with defaecatory difficulties have a greater recovery velocity of the resting anal pressure in the proximal anal canal [9].

The rectal defaecatory reflex described above is weak and is fortified by the parasympathetic defaecatory reflex in order to perform effective defaecation. The parasympathetic reflex involves the sacral segment of the spinal cord. The nerve endings in the rectum transmit signals to the spinal cord when stimulated. There is then a reflex stimulation to the descending colon, sigmoid colon and rectum via the parasympathetic nerve fibres in the pelvic nerves. The parasympathetic signals intensify the peristaltic wave and relax the internal anal sphincter. The combined parasympathetic and sympathetic reflexes can sometimes effectively empty the left side of the colon in one movement. In patients following spinal injury, the reflex may still be present and accounts for the involuntary evacuation seen in these patients.

Defaecation is voluntarily inhibited by contraction of puborectalis and the external anal sphincter; this allows defaecation to be deferred to a more convenient time. Voluntary defaecation is initiated by relaxing puborectalis and the external anal sphincter and contracting the abdominal muscles. Defaecation is best performed in the squat position (knees higher than hips, with the back straight and elbows on knees), allowing the anorectal angle to straighten aiding defaecation.

The gastrocolic reflex is another important reflex in the timing of defaecation. As the stomach distends, there is an initiation of rectal contraction and a desire to defaecate. The exact mechanism is uncertain but may be due to the effects of gastrin on the colon. In children this reflex is dominant and defaecation follows meals, whereas in adults cultural factors and individual habits determine when defaecation occurs.

Normal defaecatory patterns vary considerably between individuals with some opening their bowels 3 times a day and others every 3 days.

No standardisation exists for either the equipment or technique used for anal canal physiology. A variety of different systems have been used, and it is important to note that measurements differ depending on the system employed. Available systems include microballoons filled with air or water, microtransducers and water-perfused catheters. These systems may be hand-held or automated. Hand-held systems are withdrawn in a measured stepwise fashion with recordings made after each step (usually of 5–10 mm intervals), or a single measurement is recorded at the point of maximum rest or squeeze anal canal pressure. The automated systems withdraw the catheter at set rates and allow continuous data recording (usually at 1 mm intervals).

Each method has been validated for its repeatability and reproducibility, although individual methods are not interchangeable. The correlation between measurements made using different systems and catheters is good; the absolute values are different, and so when comparing the results of different studies, it is essential to consider the method used to obtain the pressure measurements [11].

The majority of anal physiology studies are performed using water-perfused systems. Water-perfused catheters use hydraulic capillary infusers to perfuse catheter channels, which are arranged either radially or obliquely staggered. Each catheter channel is then linked to a pressure transducer. Infusion rates of perfusate (sterile water) vary between 0.25 and 0.5 mL/min per channel. Systems need to be free from air bubbles, which may lead to inaccurate recordings, and avoid leakage of perfusate onto the perianal skin, which may lead to falsely high resting pressures due to reflex external sphincter action. Perfusion rates should remain constant, because faster rates are associated with higher resting pressures, whilst larger diameter catheters lead to greater recorded pressure.

Vector volume manometry utilises a radially arranged catheter (eight-channel catheters are most commonly used) that is withdrawn from the anal canal during rest and squeeze, and computer

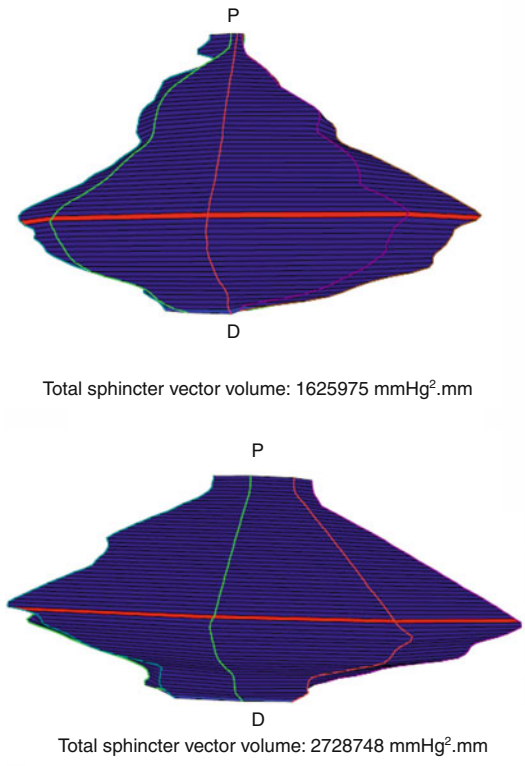

Fig. 1.2 Normal resting and squeeze vector volume profiles. The figure above shows the normal vector volume profiles at rest (*above*) and when squeezing (*below*) on the left. The axial section with the maximum pressure is identified by the *red line*, Proximal anal canal (*P*) and Distal anal canal (*D*)

software produces a three-dimensional reconstruction of the anal canal. Vector volume manometry is able to assess the anal sphincters as a three-dimensional structure (Fig. 1.2).

Vector volume manometry may differentiate between idiopathic and traumatic faecal incontinence by showing global sphincter weakness rather than a localised area of scarred sphincter indicated by an asymmetrical vectogram [12].

Significant variation exists in the results of anorectal manometry in normal asymptomatic subjects. There are no significant differences in sphincter pressures at rest between men and women, although men have significantly higher squeeze vector volume, mean maximal squeeze pressure, average squeeze pressure, squeeze high-pressure zone length (length of anal canal where the pressure is over 50 % of the maximal squeeze pressure) and length from the anal verge to peak pressure when squeezing than women. Women have a significantly more asymmetric anal canal than men on both station and automated pull through at rest and when squeezing. Anal canal pressures decline after the age of 60 years, changes most marked in women. These facts must be considered when selecting appropriate control subjects for clinical studies.

Normal mean anal canal resting tone in healthy adults is 55 to 120 mmHg. Resting tone increases in a cranial to caudal direction along the canal such that maximal resting pressure is found 6–12 mm from the anal verge. In a normal individual, the rise in pressure on maximal squeezing should be at least 50–100 % of the resting pressure [6]. It is greater in men than women (140–240 mmHg vs 110–190 mmHg) [11]. The maximum squeeze pressure is found 10–20 mm from the anal verge. Reflex contraction of the external sphincter should occur when the rectum is distended, on coughing, or with any rise in intra-abdominal pressure.

The high-pressure zone (the length of anal canal where the resting pressure is >50 % of the maximum resting pressure) is 15–25 mm in length in both men and women at rest, but the length of the high-pressure zone is longer in men when squeezing (25–35 mm vs 20–30 mm) [11].

In the assessment of patients with faecal incontinence, both resting and maximal squeeze pressures are significantly lower in patients with incontinence than in matched controls; there is, however, considerable crossover between the pressures recorded in patients and controls.

The sensation of rectal filling is measured by progressively inflating a balloon placed within the rectum with air or by an intrarectal infusion of water. Normal perception of rectal filling occurs after inflation of 20–40 mL, the sensation of the urge to defaecate occurs after 60–120 mL, and normally up to 230 mL is tolerated before discomfort occurs. The clinical use of these measurements may be limited due to the large inter- and intrasubject variation in values and the wide normal range, reducing the discriminatory value of this technique as a clinical investigation [13].

The anal canal is rich in sensory receptors, including those for pain, temperature and movement, with the somatic sensation of the anal transitional mucosa being more sensitive than that of the perianal skin. The rectum is relatively insensitive to pain, although crude sensation may be transmitted via the nervi erigentes of the parasympathetic nervous system. Normal subjects can detect anal canal changes in temperature of 0.92 °C, and normal electrical sensation for the transition zone, the most sensitive area of the anal canal, is 4 mA (2–7 mA) [14]. Temperature sensation may play a role in the discrimination of solid stool from liquid and flatus and is reduced in patients with faecal incontinence, although this is brought into question as the anal mucosa sensitivity to temperature change is not accurate enough to detect the very slight temperature gradient between the rectum and anal canal [14]. Anal mucosal electrical sensation threshold increases with age and thickness of the subepithelial layer of the anal canal. Anal canal electrical sensation is reduced in idiopathic faecal incontinence, diabetic neuropathy, descending perineum syndrome and haemorrhoids. The sampling mechanism and maintenance of faecal continence are complex multifactorial processes, as seen by the fact that the application of local anaesthetic to the sensitive anal mucosa does not lead to incontinence and in some individuals actually improves continence.

Anal Canal Imaging

The two most common modalities used to image the anal canal are magnetic resonance imaging (MRI) and ultrasound. Ultrasound is most commonly performed endoanally (anal endosonography AES), whereas MRI may be performed as a surface scanning technique or rarely endoanally (endocoil MRI). CT is unsatisfactory for the assessment of the anal canal because of the poor special resolution, largely due to volume averaging between CT slices. There are three main indications for imaging of the anal canal area: malignancy, faecal incontinence and sepsis including fistula disease.

Anal Endosonography

Two-dimensional AES utilises a transducer that rotates through 360° within a water-filled plastic cone and by using a 10-MHz transducer. The development of high-resolution three-dimensional AES constructed from a synthesis of standard two-dimensional cross-sectional images produces a digital volume that may be reviewed and used to perform measurements in any plane which yields more information on the anal sphincter complex. This provides more reliable measurements, and volume measurements can also be performed. Three-dimensional endoluminal ultrasound uses a double crystal design (frequency range from 6 to 16 MHz) encased in a cylindrical transducer shaft. The increased frequency range allows for higher resolution or deeper depth penetration.

The anal canal mucosa is generally not seen on AES. The subepithelial tissue is highly reflective and surrounded by the low reflection from the internal anal sphincter. The width of the internal sphincter increases with age: the normal width for a patient aged 55 years or younger is 2.4–2.7 mm, whereas in an older patient, the normal range is 2.8–3.4 mm [15]. As the width of the sphincter increases, it becomes progressively more reflective and more indistinct, which may be due to a relative increase in the fibroelastic content of this muscle as a consequence of ageing. Both the external anal sphincter and the longitudinal muscle are of moderate reflectivity. The intersphincteric space often returns a bright reflection. An axial section through the mid anal canal is seen in Fig. 1.3.

Another development in the use of three-dimensional AES is the use of volume rendering. This allows analysis of information inside a three-dimensional volume by digitally enhancing individual voxels. By use of the different post-processing display parameters, the volume-rendered image provides better visualisation performance when there are no large differences in the signal levels of pathologic structures compared with surrounding tissues. The anatomical structures in the pelvis, the axial and longitudinal extension of anal sphincter defects, the anatomy

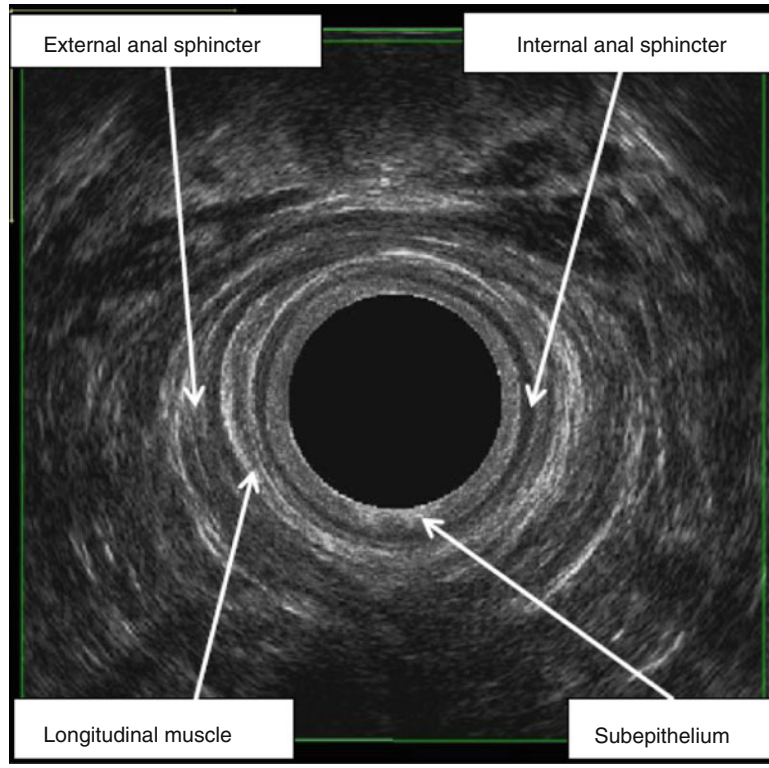

Fig. 1.3 Axial section of the normal anal canal on AES

of the fistulous tract in complex perianal sepsis and the presence of submucosal invasion in early rectal cancer may be imaged in greater detail.

Magnetic Resonance Imaging

Surface MRI is routinely performed on a 1.5 T magnet with a body coil receiver in the supine position. Endocoil MRI utilises the same 1.5 T magnet but is more invasive due to the insertion of a receiver coil in the anus. Some patients with pelvic sepsis or stenosing tumours may not be able to tolerate endocoil MRI. Endocoil MRI provides images with excellent tissue differentiation and higher special resolution (within about 4 cm) than body coil MRI. It does however have a reduced field of view compared to body coil MRI. Endocoils have either rectangular or saddle geometry and measure 6–10 cm in length and 7–12 mm in diameter. This increases to 17–19 mm after encasement in an acetal homopolymer (Delrin) former. The coil is inserted in the left lateral position and then secured with sandbags or with a purpose-built holder to avoid movement artefact.

The anal sphincters are of low signal intensity on MRI with T1-weighted images of the anal canal in contrast to the high signal intensity of the fat in the ischioanal fossa. The internal anal sphincter has an intermediate to low signal intensity on T2-weighted images and is seen as a cylindrical structure surrounding the anal canal mucosa and subepithelial tissues. With the use of gadolinium (an intravenous contrast agent used in MRI), the internal anal sphincter has a high signal intensity. The intersphincteric space has a high signal intensity, whereas the longitudinal muscle and external anal sphincter have a lower signal intensity similar to the skeletal muscle. The subepithelial tissue has a signal intensity between that of the internal and external anal sphincters.

As well as the use of contrast media, STIR (short tau inversion recovery) sequences MRI may be used to increase tumour or fistula conspicuity by suppressing the signal returned by fat [16].

However, with STIR sequence the anatomical boundaries are hard to delineate when compared to T2 images. When compared to AES, MRI is more expensive and time-consuming and may be contraindicated in certain patients (e.g. in the presence of a pacemaker or metalwork).

Primary Staging of Anal Carcinoma

AES is useful in the assessment of local disease (T stage) but due to its field of view has a limited role in assessing nodal disease. Whilst the perirectal lymph nodes are seen on AES, the iliac and inguinal nodes are beyond the range of AES. Anal tumours appear as hypoechoic lesions that may involve the anal sphincters, with invasion through the sphincter layers. Lymph nodes are hypoechoic, and enlarged lymph nodes, over 10 mm, in the mesorectum may suggest lymph node metastasis. It is difficult to distinguish between metastasis and reactive lymph nodes; malignant nodes may be more circular, lose the normal hilar structure and be of similar reflectivity to the primary tumour. In the staging of small superficial tumours of the anal canal, AES has been shown to be superior to MRI [17]. Three-dimensional AES is superior to standard AES in both the assessment of the primary anal tumour and the assessment of perirectal lymph nodes [18].

MRI provides clear delineation of the anatomical boundaries in the anal canal and is used to plan patient's radiotherapy. On MRI anal tumours have an isointense signal intensity on T1-weighted scans and intermediate signal intensity on T2-weighted scans. Tumour extension is identified by the loss of the clear anatomical definition and ill-defined indeterminate signal intensity. Metastatic lymph nodes on MRI have a similar signal to that of the primary tumour on either T1- or T2-weighted imaging. Perirectal nodes over 5 mm in diameter and inguinal or pelvic side wall nodes over 10 mm in maximum suggest metastasis. Though again it is difficult to distinguish between metastasis and reactive nodes and to assess microscopic nodal involvement.

In the evaluation of anal tumours following treatment, AES may be used to assess the response, but oedema and scaring are difficult to distinguish from persistent or recurrent disease. Three-dimensional ultrasound has again been shown to be superior to standard ultrasound in the assessment of recurrent disease. On MRI the size of the tumour and signal intensity is reduced following response to chemoradiotherapy. The reduction in tumour size is most evident 6 months after treatment when the surrounding inflammation has settled. As with AES follow-up with MRI can lead to over or under staging due to the appearances of tumour fibrosis and reactive lymphadenopathy. Recurrent disease on MRI has a signal intensity similar to that of the primary disease [19].

Imaging of Anal Fistula

On AES it is rare to visualise the internal opening (submucosal defect) but can usually be inferred as the point where the tract fuses with the internal sphincter. A fistula tract is seen on AES as a hypoechoic band tracking through the anal canal. Gas or foreign material (such as a seton suture) within the tract will appear as a bright reflection. An abscess cavity or further extension of the fistula tract will again be hypoechoic. Figure 1.4 shows the AES appearances of a high transsphincteric fistula controlled with a seton.

On MRI sepsis appears as areas of very high signal intensity. This can be enhanced using the intravenous contrast agent gadolinium or with the use of STIR (short tau inversion recovery) sequence [16]. STIR sequence has an increased sensitivity in detecting the internal opening compared to gadolinium-enhanced scans. Figure 1.5 shows the axial appearances of a transsphincteric anal fistula as seen on STIR sequence MRI.

Both AES and MRI have been shown to be accurate for the definition of the anatomy of anal sepsis. The correct classification of perianal fistulae using clinical examination alone is 61 %, compared with endosonography 81 % and MR imaging 90 %. The ability of AES and MRI to predict the site of the internal opening accurately is comparable 91 % for AES versus 97 % for MRI [20]. AES has also been shown to be accurate for the definition of the anatomy of complex

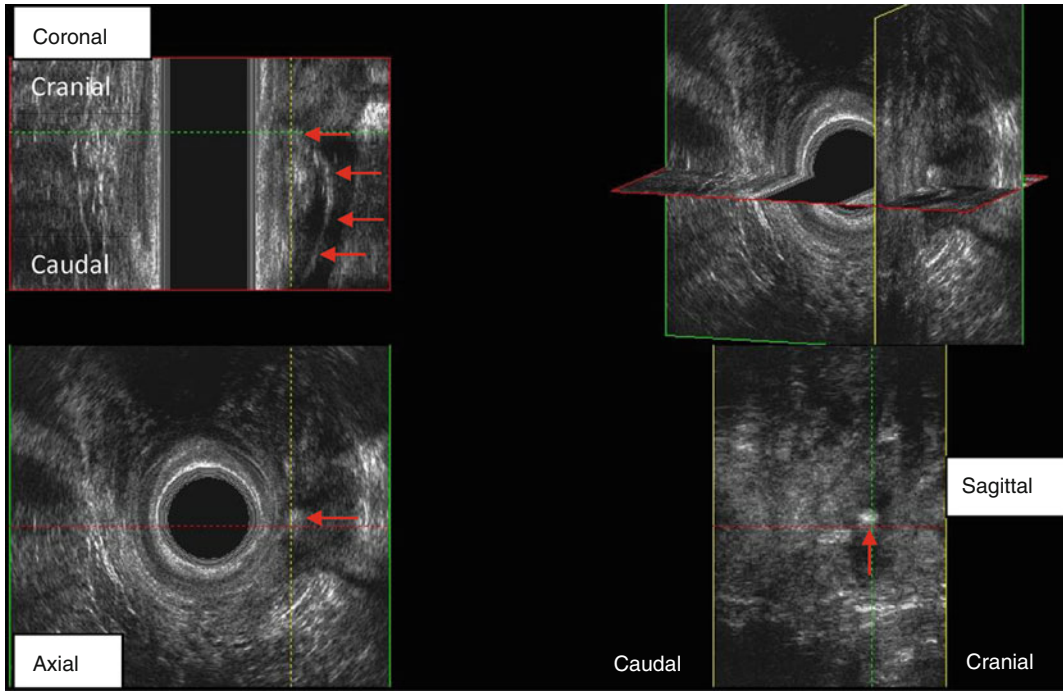

Fig. 1.4 Anal fistula controlled with a seton. The figure above shows the AES appearances of a high transsphincteric fistula, with an internal opening cranial to the external anal sphincter at 3 o'clock, controlled with a seton (*marked with red arrows*). The seton is hyperechoic and can be seen surrounded by the hypoechoic fistula tract

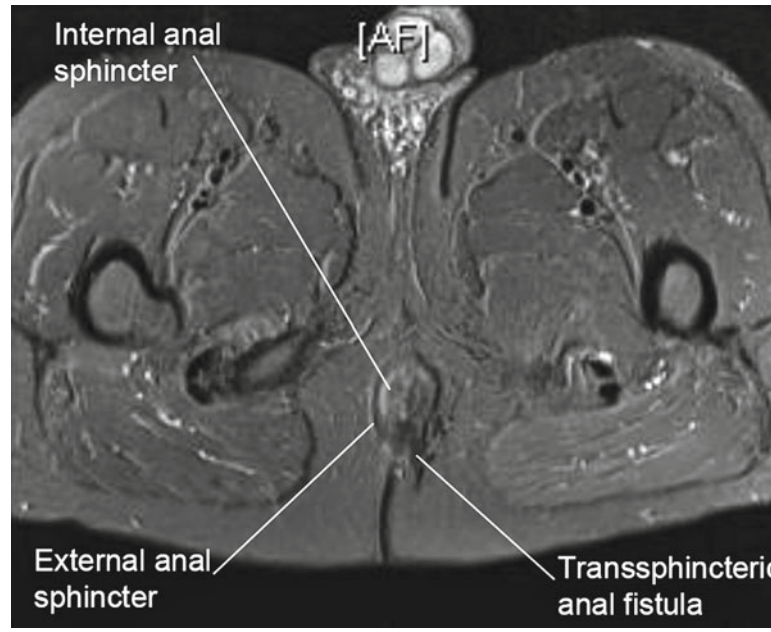

Fig. 1.5 Axial view of a transsphincteric fistula as seen on STIR sequence MRI. The figure above shows an axial section of a STIR sequence MRI. A transsphincteric fistula is seen, with an internal opening at the 6 o'clock position and an external opening at the 5 o'clock position

anal sepsis, especially horseshoe collections and in the assessment of sphincter damage caused by chronic sepsis. AES is however less accurate in the assessment of suprasphincteric sepsis, and it is often difficult to differentiate between supralevator and infralevator collections, leading to inaccuracy in up to 20 % of cases [21]. The diagnostic accuracy of AES is increased with the use of hydrogen peroxide injected into fistulous tracts to act as a contrast medium. Three-dimensional AES has again been shown to be superior to two-dimensional AES in the accurate diagnosis of perianal fistula [22].

Imaging in Faecal Incontinence

AES and MRI can be used to assess patients with faecal incontinence. The accuracy of AES in the diagnosis and assessment of the extent of external sphincter injury has been validated in comparison studies with EMG and findings at surgery. AES has been used to identify patients with sphincter defects that were initially believed to have idiopathic faecal incontinence [23]. AES has also shown that a much higher proportion of women sustain sphincter damage during childbirth than is suspected by clinical assessment alone. AES is superior to anal physiology assessment in the differentiation between patients with idiopathic faecal incontinence and those with a sphincter defects. MRI is also used to assess patients with faecal incontinence and has been validated with surgical confirmation of the presence and extent of sphincter defects. Three-dimensional AES and endoanal MRI can also be used in the assessment of anal sphincter atrophy. Both modalities are comparable in detecting atrophy and defects to the external anal sphincter, though the correlation between the two for measurement of sphincter thickness and length is poor.

There are certain features relevant in the clinical assessment of men who have sex with men. Two studies have assessed the effects of anoreceptive intercourse on sphincter function. The first study identified a significant increase in faecal incontinence symptoms following anoreceptive intercourse. There was a decrease in the maximum resting following anoreceptive intercourse. The maximum squeeze pressure was reduced following anoreceptive intercourse only in those faecal incontinence symptoms. The second study concluded that passive anoreceptive intercourse was associated with decreased resting pressures, but there was no change in anal squeeze pressure. There was also no internal or external anal sphincter defects on AES, and none of those studied had any faecal incontinence symptoms [24]. It was suggested the reduced resting pressure was due to better relaxation during the examination [24].

There is also an increased risk of sexually transmitted disease with human papillomavirus in particular, which must be taken into account on examination. The prevalence of human papillomavirus infection in the anal canal is higher among sexually active homosexual men (40–5 0 % in HIV-negative men and 90 % in human immunodeficiency virus-positive men). The risk is greater with a greater number of lifetime sexual partners. Most human papillomavirus infections are transient, but they can lead to anal warts and preneoplastic and malignant anal lesions. The risk of human papillomavirus infection is 44 times higher in homosexual men than among the general population. The risk of anal cancer is approximately 60 times higher than that of the general population [25]. As there is an increased risk, it has been suggested that clinicians should be regularly performing digital rectal examination in those at high risk of anal cancer.

Summary

A comprehensive understanding and assessment of the anal canal play a vital role in the correct diagnosis and management of patients with anal canal disorders. Imaging of the anal canal may be performed using either AES or MRI, for evaluation of anal cancer, anal fistula and anal incontinence. Staging of early anal cancer is superior with AES though MRI is superior in the assessment of distant lymph node metastasis. MRI is slightly superior in the assessment of anal sepsis although AES does provide a useful alternative.

AES is the gold standard in assessing anal sphincter integrity following obstetric injury. A wide variety of tests are available to assess the physiology of the anal canal. Physiological assessment gives an objective measure in patients with both faecal incontinence and difficult defaecation. The understanding of the anal canal anatomy and physiology is vital in the correct management of patients with anal disorders.

References

1. Parks AG. Modern Concepts of the Anatomy of the Ano-Rectal Region. Postgrad Med J. 1958; 34(393):360–66.
2. Hollinshead WH. Embryology and anatomy of the anal canal and rectum. Dis Colon Rectum. 1962; 5:18–22.
3. Goligher JC, Leacock AG, Brossy JJ. The surgical anatomy of the anal canal. Br J Surg. 1955;43:51–61.
4. Thompson P. On the Levator Ani, or Ischio-Anal Muscle of Ungulates, with Special Reference to its Morphology. J Anat Physiol. 1899;33(3):423–33.
5. Oh C, Kark AE. Anatomy of the external anal sphincter. Br J Surg. 1972;59(9):717–23.
6. Williams AB, Cheetham MJ, Bartram CI, Halligan S, Kmiot WA, Nicholls RJ. Gender differences in the longitudinal pressure profile of the anal canal related to anatomical structure as demonstrated on three-dimensional anal endosonography. Br J Surg. 2000;87:1674–9.
7. Lestar B, Penninckx F, Kerremans R. The composition of anal basal pressure. An in vivo and in vitro study in man. Int J Colorectal Dis. 1989;4:118–22.
8. Liu J, Guaderrama N, Nager CW, Pretorius DH, Master S, Mittal RK. Functional correlates of anal canal anatomy: puborectalis muscle and anal canal pressure. Am J Gastroenterol. 2006;101(5):1092–7.
9. Kaur G, Gardiner A, Duthie GS. Rectoanal reflex parameters in incontinence and constipation. Dis Colon Rectum. 2002;45(7):928–33.
10. Miller R, Bartolo DC, Cervero F, Mortensen NJ. Anorectal sampling: a comparison of normal and incontinent patients. Br J Surg. 1988;75:44–7.
11. Schizas AM, Emmanuel AV, Williams AB. Vector volume manometry – methods and normal values. Neurogastroenterol Motil. 2011;23(9):886–e393.
12. Braun JC, Treutner KH, Dreuw B, Klimaszewski M, Schumpelick V. Vectormanometry for differential diagnosis of fecal incontinence. Dis Colon Rectum. 1994;37(10):989–96.
13. Kendall GPN, Thompson DG, Day SJ, Lennard-Jones JE. Inter- and intraindividual variation in pressure-volume relations of the rectum in normal subjects and patients with irritable bowel syndrome. Gut. 1990;31:1062–8.
14. Felt-Bersma RJ, Poen AC, Cuesta MA, Meuwissen SG. Anal sensitivity test: what does it measure and do we need it? Cause or derivative of anorectal complaints. Dis Colon Rectum. 1997;40(7):811–6.
15. Burnett SJ, Bartram CI. Endosonographic variations in the normal internal anal sphincter. Int J Colorectal Dis. 1991;6(1):2–4.
16. Buchanan GN, Halligan S, Williams AB, et al. Magnetic resonance imaging for primary fistula in ano. Br J Surg. 2003;90(7):877–81.
17. Otto SD, Lee L, Buhr HJ, Frericks B, Hocht S, Kroesen AJ. Staging anal cancer: prospective comparison of transanal endoscopic ultrasound and magnetic resonance imaging. J Gastrointest Surg. 2009; 13(7):1292–8.
18. Christensen AF, Nielsen MB, Engelholm SA, Roed H, Svendsen LB, Christensen H. Three-dimensional anal endosonography may improve staging of anal cancer compared with two-dimensional endosonography. Dis Colon Rectum. 2004;47(3):341–5.
19. Roach SC, Hulse PA, Moulding FJ, Wilson R, Carrington BM. Magnetic resonance imaging of anal cancer. Clin Radiol. 2005;60(10):1111–9.
20. Buchanan GN, Halligan S, Bartram CI, Williams AB, Tarroni D, Cohen CR. Clinical examination, endosonography, and MR imaging in preoperative assessment of fistula in ano: comparison with outcome-based reference standard. Radiology. 2004; 233(3):674–81.
21. Deen KI, Williams JG, Hutchinson R, Keighley MR, Kumar D. Fistulas in ano: endoanal ultrasonographic assessment assists decision making for surgery. Gut. 1994;35(3):391–4.
22. Santoro GA, Fortling B. The advantages of volume rendering in three-dimensional endosonography of the anorectum. Dis Colon Rectum. 2007;50(3): 359–68.
23. Burnett SJD, Spence-Jones C, Speakman CT, Kamm M, Hudson CN, Bartram CI. Unsuspected sphincter damage following childbirth revealed by anal endosonography. Br J Radiol. 1991;64:225–7.
24. Chun AB, Rose S, Mitrani C, Silvestre AJ, Wald A. Anal sphincter structure and function in homosexual males engaging in anoreceptive intercourse. Am J Gastroenterol. 1997;92(3):465–8.
25. Goldstone S, Palefsky JM, Giuliano AR, et al. Prevalence of and risk factors for human papillomavirus (HPV) infection among HIV-seronegative men who have sex with men. J Infect Dis. 2011;203(1): 66–74.

Anorectal Physiology

2

Jonathan Gosling and Anton Emmanuel

Introduction

Disorders of the pelvic floor are common and cause considerable morbidity. It is important to identify which patients need which intervention and as importantly which patients will not benefit from intervention. This in turn depends on accurate clinical and physiological assessment as well as a multidisciplinary and multimodal approach to treatment. The latter is especially important since many of these patients have a global pelvic floor dysfunction which requires a holistic approach to the pelvic floor rather than dividing it into anterior, middle and posterior compartments. This chapter is divided into three sections:

1. Clinical assessment: history and examination
2. Standard physiological measurements
3. Advanced physiological measurements

Clinical Assessment

History

Constipation

Constipation is a common symptom in gastroenterological and surgical clinics. Prevalence depends on the definition used, but a telephone interview-based study conducted in the USA which included over 10,000 individuals found the prevalence to be 14.7 % [1].

Definition

Constipation is an umbrella term used to describe a group of individual symptoms that commonly coexist. These symptoms include decreased frequency of defaecation, difficulty in evacuating (requiring excessive straining, anal or vaginal digitation), feeling of incomplete emptying (which often results in repeated unsuccessful attempts to evacuate) and hard stool consistency. The most frequently used definition and one that is commonly used in research studies is the Rome III criteria for functional constipation which are outlined in Table 2.1 [2].

Table 2.1 Rome III criteria for constipation [1]

1. Must have at least 2 of the following:
(a) Straining during at least 25 % of defaecations
(b) Lumpy or hard stools in at least 25 % of defaecations
(c) Sensation of incomplete evacuation for at least 25 % of defaecations
(d) Sensation of anorectal obstruction/blockage for at least 25 % of defaecations
(e) Manual manoeuvres to facilitate at least 25 % of defaecations
(f) Fewer than three defaecations per week
2. Loose stools are rarely present without the use of laxatives
3. Insufficient criteria for irritable bowel syndrome

Longstreth et al. [2]
Criteria fulfilled for the last 3 months with symptom onset at least 6 months prior to diagnosis

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Classification

Constipation can be classified into primary and secondary constipation. Secondary causes include colonic and small bowel obstruction, hypothyroidism, hypercalcaemia and medications in particular opioid analgesics. Secondary causes should be excluded before undertaking investigations for primary constipation. Primary constipation is classified pathophysiologically into slow-transit constipation (STC), constipation-predominant irritable bowel syndrome, evacuation dysfunction or a combination of these pathologies [3]. Irritable bowel syndrome may be associated with symptoms of constipation, but abdominal pain must be present for constipation to be ascribed to this particular functional condition. Patients with neither slow-transit constipation, evacuation difficulty nor IBS can be referred to as having functional constipation. Some use functional constipation to refer to STC or any cause of primary constipation, and its ambiguity means it should be avoided if possible. Difficulty in evacuating or obstructive defaecation can be caused by anatomical abnormalities (rectocele, enterocoele, rectal prolapse and intussusception) or by a functional disorder. Such functional problems may relate to rectal hyposensitivity [4] or pelvic floor dyssynergia [5] for which there are multiple names including anismus, paradoxical puborectalis contraction and rectoanal dyssynergia.

Differentiating Causes Using History

Although there is considerable overlap between the various subtypes of primary constipation, careful history and investigations can help subclassify patients in order to guide subsequent treatment.

Slow-transit constipation may be differentiated from other types of constipation as it typically starts in teenager or young adult phase and is more common in women. It results in an infrequent urge to defaecate and a hard stool consistency [6]. Patients with an evacuation disorder report a constellation of symptoms which help differentiate this group from the slow-transit group. Although not all universally present, they include excessive straining, the feeling of incomplete evacuation requiring multiple evacuatory attempts, pelvic pain, anal or vaginal digitation to assist defaecation, assuming an unusual position during defaecation and application of pressure to the perineum, anal margin or buttocks.

Scoring and Questionnaires

Questionnaires for constipation can be grouped into those assessing overall severity of constipation (such as the Wexner constipation score [7]), those assessing a specific aspect of constipation (such as the Obstructive Defecation Score) and those which attempt to aid differentiating patients into the various subgroups (such as the Constipation Severity Instrument [8]). Questionnaires are generally used in the context of research rather than aiding in clinical decision making.

Incontinence

Definition

Faecal incontinence is defined as the involuntary passage of faecal matter through the anal canal. It is a common condition with a general population incidence of approximately 2 % [9] although this increases to 26 % of women over the age of 50 [10]. Social stigma means that it is an underreported condition, strongly associated with reduced quality of life and self-confidence. It is one of the leading causes of nursing home placement [11]. As a result, presentation may be different to that of other conditions and may be discovered on examination. Empathic questioning style will enable the patient to feel comfortable revealing the severity of the problem. Presenting symptoms include urgency and urge incontinence (suggestive of external anal sphincter dysfunction) or passive leakage of liquid or solid stools (typically suggesting internal anal sphincter dysfunction).

Classification

The continence mechanism relies on a number of factors: the local ones include internal and external anal sphincter function, rectal reservoir function and anorectal sensation, but it is also important to consider the upstream contributors

Table 2.2 Classification of faecal incontinence

Sphincteric	Sphincter injury	Obstetric	
		Surgical/iatrogenic	
		Trauma	
	Neurological	Pudendal neuropathy	Obstetric
			Chronic straining
		Multiple sclerosis	
		Diabetes mellitus	
		Spinal cord injury	
		Cauda equina syndrome	
Suprasphincteric	Reduced rectal compliance	Radiotherapy	
		Proctitis	
		Spinal injury	
	Diarrhoea	Side effect of medications	
		Laxative abuse	
		Abnormal colonic function	
		Abnormal small bowel function	
	Rectal prolapse		
	Rectal impaction		
Higher cortical function	Dementia		
	Learning difficulties		

such as colonic and small bowel function, adequate response of somatic muscles to raised rectal pressure and normal higher cognitive function. Incontinence results from impairment to one or a number of these continence mechanisms. Table 2.2 gives a broad classification system which is based around these continence mechanisms. Although many patients commonly do not fall neatly into any one of these categories, it is useful to have this structure in mind.

Differentiating Causes Using History

Obstetric injury is the leading cause of faecal incontinence, and as such a detailed obstetric history should be elicited. As an indication of prevalence following child birth, 13 % of women develop urgency or incontinence following their first delivery [12]. Symptoms may be transient following child birth with two thirds of those who experience flatus incontinence recovering fully within a year, although approximately 4 % have persistent incontinence following 'uncomplicated' vaginal delivery [13].

The onset of symptoms may be some time following obstetric injury with patients presenting in the 50s or 60s. Explanations for the latency include failure of compensatory mechanisms and progressive pudendal injury from perineal descent. The delayed onset means sphincter injury should be sought in women of any age who have had a vaginal delivery presenting with faecal incontinence. Indications in the history that women are of high risk of having a sphincter injury include a birth weight of greater than 4 kg [14], prolonged second stage greater than 4 h (odds ratio of 1.33 for sustaining a third- or fourth-degree tear [15]), previous sphincter damage (five to seven times more likely to sustain a repeated obstetric injury [16]) and instrumental delivery (<13–81 % of forceps deliveries result in a third-degree tear, and nearly two thirds go on to having incontinence [17, 18]). There is no evidence for the protective effect of an episiotomy, and it could even increase the risk of sphincter injury, although mediolateral episiotomy is less harmful than midline [19].

Caesarean section performed electively or at an early stage of labour reduces the risk of obstetric tears. A common clinical question from patients and obstetricians is regarding advice on mode of future deliveries. Although it is not recommended that primary elective caesarean

section should be offered to women to preserve continence, a case can be made for those with high risk of sphincter injury including those with a previous sphincter injury or previous symptoms of incontinence to be offered elective Caesarean section [20, 21].

Although obstetric injury is the most common cause of faecal incontinence, even in patients with a good history of obstetric injury, it is important, especially in elderly patients, to exclude the presence of faecal impaction or rectal prolapse. In addition colonic disease should be excluded in all patients presenting with faecal incontinence associated with a change in stool consistency or frequency of defaecation.

In addition to obstetric trauma, anal sphincter damage can occur during surgery for haemorrhoids, anal fistula or transanal resections. Accidental injury and nonconsensual anal intercourse are also associated with sphincter disruption. Faecal incontinence in the presence of normal sphincter anatomy, previously termed idiopathic incontinence, is frequently secondary to pudendal nerve injury that occurred during labour or from stretching during chronic excessive straining in perineal descent syndrome. Diseases affecting the central and peripheral nervous systems such as multiple sclerosis, stroke and diabetes are commonly complicated with faecal incontinence. Illnesses causing diarrhoea can present with or be complicated by faecal incontinence, and these include inflammatory bowel disease, laxative abuse and short gut syndrome. Investigations in these cases should focus on the cause of diarrhoea before undergoing physiological testing of the continence mechanism. Pelvic irradiation can cause faecal incontinence by reducing rectal compliance and impairing sphincter tone.

Scoring Systems

Quantification of the symptom has been attempted through a variety of scoring systems: the Pescatori [22], the Wexner [23], the American Medical Systems Score and the Vaizey Score [24]. All have been shown to have a good correlation with overall clinical impression of severity. Incontinence scores are normally used in the context of clinical trials rather than part of routine clinical practice.

Examination

Careful examination can help elicit the severity and cause of faecal incontinence and evacuatory difficulty.

Inspection

The severity of passive incontinence can be assessed by the degree of soiling on the perianal skin, underwear or pad. Perianal excoriation and erythema is an indication that passive incontinence is causing pruritus ani. On occasion symptoms of passive loss of faecal matter are not due to faecal incontinence but due to perianal fistulas, and an external fistula opening can be found. The anocutaneous reflex can be elicited by stimulating the perianal skin with either a pin prick or light touch and contraction of the external anal sphincter; the so-called anal wink should occur. The pudendal nerve transmits both the afferent and efferent signals, and the synapse occurs in the caudal spinal cord.

DRE

Digital rectal examination should be performed after anal manometry to maximise the precision and accuracy of these physiological parameters. Resting tone squeeze pressure can be estimated and, although not as accurate or reproducible as manometric measurements, provides clinicians without direct access to this equipment an estimation of internal and external sphincter function. The integrity of the anal sphincters can be examined by rotating the examining finger through 360° checking for a complete anorectal ring. Defects of less than 90° are difficult to identify clinically [25]. The anal sphincter is shorter anteriorly in women, which can be misinterpreted as an anterior sphincter defect. Application of pressure to the rectovaginal septum can reveal the presence of a rectocele [26]. The size of the weakness can be estimated as localised or diffuse. The anal canal pressure can be estimated digitally during straining or attempted defaecation. The absence of an increase in anal canal pressure under these circumstances suggests the absence of anismus [27].

Attempted Defaecation and Balloon Expulsion

Patients with symptoms of prolapse and elderly patients with a history of constipation who present with passive incontinence should be thoroughly examined for the presence of a full-thickness rectal prolapse. They should be asked to strain as heavily as they would to pass stools whilst on a toilet or commode and given enough time to reproduce the prolapsing lump before examination.

Expulsion of a 60 ml water-filled balloon on a commode or toilet can be used in the assessment of constipated patients. The ability to expel the balloon within 1 min may be a useful tool in demonstrating the absence of pelvic floor dyssynergia [28].

Standard Physiology

Introduction

The tools that are at the disposal of anorectal physiology laboratories in the functional assessment of faecal incontinence and evacuatory disorders aim to measure function and determine the cause of the presenting condition in order to guide treatment and monitor progress. Our understanding of anorectal physiology and pathophysiology is limited by the precision and accuracy of these tests. They include anal manometry, balloon distension studies, electrical anal and rectal mucosal studies, barostat studies, impedance planimetry and investigations aimed at ascertaining pudendal nerve function including pudendal nerve terminal motor latencies and single-fibre electromyographs. The standard physiology tests used in routine clinical practice are anal canal manometry and rectal and anal sensory measurements. These will be discussed in this section.

Manometry

Manometric assessment is routinely performed in many centres as part of the work-up for faecal incontinence to assess the mechanical strength of the anal sphincters. The measurements provided are limited by a degree of imprecision and inaccuracy. Identifying aetiology by manometry alone is not usually possible. Therefore in clinical practice, manometry results are interpreted with caution and guide treatment only in conjunction with clinical assessment and other investigations, in particular modern imaging techniques. Endoanal ultrasound images the IAS with a high level of precision and accuracy. It provides information on the thickness of IAS and identifies any sphincter defects.

Methodology

Equipment

There are various methods of measuring the pressure exerted by the anal sphincters including water-perfused, solid-state and microballoon systems.

Water-perfused systems use a 4–5 mm diameter disposable catheter with multiple narrow channels (<0.5 mm) running longitudinally along its length. The flow of water is driven by pressure of 1,000 mBar. The rate of flow through the catheter is limited by a flow resistor to give a flow rate of 0.15–0.5 ml/min, depending on the number of channels used. The pressure exerted on the opening of the channel is transmitted via the column of water to a pressure transducer in the central control unit. Higher flow rates provide a greater response rate (the maximum increase in pressure per unit time) but deliver more water into the rectum which can be troublesome in long studies and in systems with a large number of channels.

Solid-state systems have pressure transducers within the catheter. They are generally more expensive than water-perfused systems. As the catheter is not disposable and contains the pressure transducers, they are potentially less robust. They have a greater response rate than water-perfused systems.

Microballoon systems transmit pressure within a small intra-anal balloon to a remote pressure transducer via a closed column of water. This gives an overall measure of anal canal pressure rather than at specific points. It cannot therefore provide a measure of radial symmetry.

The channel openings in water-perfused catheters and the pressure transducers in solid-state catheters can be placed in different configurations. Typically they are arranged radially with four to eight points of measurement. They can also be arranged in a spiral configuration allowing simultaneous pressure measurements at different levels of the anal canal.

Protocol

When using a catheter with radially arranged pressure transducers, the pressure along the length of the canal is measured by either station pull through or continuous pull through. Station pull through involves inserting the catheter to 6 cm from the anal verge and withdrawing the catheter at 5–10 mm intervals and measuring for 1–5 min at each 'station'. Continuous pull through involves withdrawing the catheter at a set speed by hand or by a mechanical puller. Continuous pull through technique results in higher pressures possibly through movement-induced recruitment of the external anal sphincter or simply movement artefact.

Manoeuvres

Pressures are typically measured during rest, squeeze, endurance squeeze, cough and distension of a rectal balloon to assess the rectoanal inhibitory reflex. In addition for patients with suspected animus, anal canal pressures can be measured during straining.

Using Normal Values

Absolute pressure values measured differ depending on the sensor technology type, pressure sensor arrangement, catheter diameter and method of catheter withdrawal. There is no consensus on which of the above variables are superior, and published normal values are only valid when using the same equipment and protocol. As such the recommendation is for each physiology department to generate its own normal values from healthy volunteers from the local population. A further complicating factor is that there is a large variation within a population especially between people of different age, sex and parity. Specifically with regard to changes in resting pressure with age, the IAS thickness normally increases with age, and thinning of the muscle is associated with lower resting pressures. This can be demonstrated histologically with muscle atrophy and necrosis and extensive deposition of collagen. Ideally then, normal values should be stratified to account for different patient groups.

Interpretation of Results

Resting Pressure

The pressure is calculated for all levels of the anal canal during the resting period. If a radially arranged catheter is used, the mean pressure is used. The highest intra-anal pressure is the resting pressure and is expressed as cmH_2O or mmHg relative to atmospheric pressure. The functional length of the anal canal is calculated by the point (either station or distance depending on whether station pull through or continuous pull through is used) that the pressure rises significantly above rectal pressure. There is significant longitudinal variation in anal canal pressure, and the region of the anal canal that has a pressure above 50 % of the maximum pressure is defined as the high-pressure zone. There is also significant radial variation in anal canal pressures. Proximally the pressure is higher posteriorly owing to the action of the puborectalis sling. In the mid anal canal and high-pressure zone, the pressures are at their most symmetrical. Distally the pressures are higher anteriorly resulting from the external sphincter tonal activity. Within the limitations specified above, the normal range for resting pressure is approximately 60–120 cmH_2O.

The smooth muscle of the internal anal sphincter (IAS) and the striated muscle of the external anal sphincter (EAS) overlap and surround the anal canal where the contraction of the individual muscles summate to create the positive intra-anal pressure. The IAS exhibits continuous tonic activity and is responsible for 55–85 % of the resting anal canal pressure [29–31]. Its contribution to resting tone is variable along the length of the anal canal with the proximal two thirds being more reliant on IAS tone to maintain adequate resting pressures. Structurally the IAS is the anal canal's equivalent of the circular muscle in the rectum. The IAS is 1–1.5 cm long with its caudal margin

demarcated anatomically by the intersphincteric groove, beyond which the EAS continues in isolation. The EAS has constant tonic activity contributing to the resting anal canal pressure [32]. The tonic activity within the EAS disappears in spinal injury patients without significant afferent input from the anal canal [33]. The tonic activity within the EAS therefore relies upon input from supraspinal centres. Histologically the EAS is composed of largely type 1 muscle fibres characteristic of muscles with constant tonic activity capable of sustained contraction utilising oxidative metabolism.

The pressure generated by the resting tone of the IAS maintains a positive anorectal pressure gradient. The extent to which the IAS is needed to generate the resting pressure is not known, as internal anal sphincterotomies can be performed for anal fissures with usually only minor disturbances in continence, providing a full-length division of the sphincter is avoided. The interactions between the vascular anal cushions and the IAS may well be vital in the fine control of continence. The IAS cannot close the anal canal completely and requires these anal cushions to fill a gap of at least 7–8 mm in diameter [34].

The tonic activity within the IAS is variable and slow, and ultraslow waves have been identified. Slow waves occur with a frequency of 10–20/min [35]. They are more frequent in the distal anal canal with the proposed function of moving anal canal contents proximally into the rectum. Ultraslow waves are less than 2/min and are not always present. They are likely to be a result of rectal stimulation inducing IAS relaxation.

The IAS has intrinsic and extrinsic autonomic innervation which modulates its activity. There is conflicting evidence on the response of the IAS to parasympathetic and sympathetic activity and which neurotransmitters and receptors mediate these responses. Lubowski et al. induced IAS relaxation by stimulating the presacral nerves and also administering alpha- and beta-blocking agents which indicates that sympathetic outflow acting via alpha-adrenoreceptors may cause an inhibitory response in the IAS [36]. However, Parks et al. found that in vitro the IAS has a contractile response to noradrenaline [37], and Frenkner et al. used spinal anaesthesia at different levels to demonstrate that there is a tonic excitatory sympathetic outflow to the IAS [38]. Nitric oxide and other non-adrenergic and non-cholinergic (NANC) neurotransmitters are also likely to be key inhibitory mediators [39, 40].

The most common cause of a low resting pressure is an IAS defect caused by obstetric or surgical trauma, with the defect usually demonstrable on endoanal ultrasound. Surgical trauma includes accidental injury during haemorrhoidectomy, anal stretch procedures, fistula surgery, sphincterotomies for anal fissures and damage during sphincter saving rectal resection surgery via a number of mechanisms including inadvertent dilatation. The reduced resting pressures result in passive faecal incontinence. External anal sphincter injuries usually accompany obstetric IAS injury and urgency, and urge incontinence may be the primary presenting symptom in this group. IAS injury can rarely occur in isolation from EAS injury in obstetric injury due to the shearing forces generated during labour. In addition to IAS defects, IAS atrophy can be identified on endoanal ultrasound.

Despite increasing understanding of the factors involved in the modulation of IAS tone, therapeutic pharmacological intervention to increase IAS tone is limited. Loperamide has been found to increase the volume required to induce IAS relaxation. This effect is inhibited by naloxone suggesting this response is mediated by opioid receptors. There is also evidence that loperamide increases resting pressures although the mechanism of action is not understood [41].

With sphincter preserving surgery becoming more common, attention has been directed at understanding the pathophysiology of continence disturbance that can complicate this type of surgery. The resting tone is commonly reduced, and this is thought to be caused by disturbance in the extrinsic autonomic nerve supply, direct trauma due to stretch and excision of the internal anal haemorrhoidal complex. Stapled rather than hand-sewn anastomosis more completely preserves IAS tone [42].

Elevated resting pressures are associated with the presence of anal fissures. The notion that

impaired vascular supply caused by raised internal anal sphincter tone causes anal fissures is supported by the findings of Doppler laser flowmetry [43]. The posterior midline, the most common site of an anal fissure, has been shown to receive the lowest flow rate of the four segments. In addition, there is a direct relationship between resting anal pressure and anodermal blood flow, with decreased resting pressures resulting in increased blood flow. Glyceryl trinitrate 0.2 % cream reduces anal canal pressure by around a quarter [44], and internal sphincterotomy results in a 26–50 % reduction in anal canal pressures [45]. GTN cream also causes anodermal mucosal vasodilation. Topical 2 % diltiazem is as effective but is not complicated with headaches which are a common side effect of GTN cream [46].

Squeeze Pressure

Although the EAS contributes to the resting pressure, the specific function of the EAS can be assessed during the squeeze, cough and rectal balloon inflation manoeuvres. The pressure increment above resting pressures during these manoeuvres is a direct representation of EAS function. The normal range is approximately above 50 cmH_2O.

The external anal sphincter and the striated muscles of the pelvic floor form an integrated functional unit which contribute to the resting anal pressure but also contract to maintain a positive anorectal pressure in response to raised rectal pressure generated by raised intra-abdominal pressure or rectal filling.

The external anal sphincter (EAS), levator ani and puborectalis form a continuous funnel of striated muscle which comes to surround the internal anal sphincter and the anal canal. Puborectalis has been found to be separated from the external anal sphincter by a distinct fascia although its histological resemblance to the EAS suggests it serves a similar function. The external anal sphincter itself can be subdivided into deep, superficial and subcutaneous sections.

The most common cause of external anal sphincter dysfunction is obstetric injury. Grade 3a involves less than 50 % and grade 3b involves greater than 50 % of the EAS. The injury causes a tear in the anterior portion of the muscles which is typically repaired primarily using an end-to-end or overlapping technique. Persisting sphincter defects cause dysfunction due to the mechanical disadvantage of an absent continuous muscular ring.

There is much interest into the pathophysiology of incontinence in patients without a structural defect of the external anal sphincter, previously termed idiopathic faecal incontinence. The concept of the dysfunction being neuropathic in nature was first described by Sir Alan Parks [47]. On histological examination of the external anal sphincter and puborectalis in patients with idiopathic faecal incontinence, they found marked myopathic changes consistent with denervation and reinnervation in addition to loss of myelinated axons and endoneural fibrosis of the intramuscular nerve fascicles. The changes were found to be most marked in the EAS and relatively spared in levator ani and puborectalis. The findings of these histological studies are consistent with other methods of diagnosing neuropathy including elongation of motor unit potential duration and increased nerve fibre density in single-fibre electromyography studies [48]. The EAS is innervated by branches of the pudendal nerve, whereas the relatively spared puborectalis is innervated directly by the S3 and S4 nerve roots. Pudendal nerve damage can occur by stretching during childbirth, stretching due to pelvic floor descent or entrapment. Entrapment can occur when the nerve emerges from between coccygeus and piriformis, at the greater or lesser sciatic notches or in the pudendal (Alcock's) canal. Pudendal nerve damage therefore seems likely to be a contributory factor in patients with incontinence without structural sphincter defects. As such idiopathic faecal incontinence is now commonly termed neuropathic faecal incontinence in contrast to myopathic faecal incontinence which occurs with sphincter defects. In fact there is considerable overlap in these two types with the majority of patients with obstetric EAS defects having evidence of pudendal neuropathy with increased muscle fibre density on single-fibre EMG studies [49]. This reflects their common obstetric aetiology.

Endurance Squeeze

The ability of the external sphincter to generate an elevated pressure over a prolonged period is an important characteristic. It allows a positive anorectal pressure gradient to be maintained following rectal filling to allow time for the rectal pressure to normalise as it undergoes the process of receptive relaxation. The predominance of type 1 skeletal muscle fibres makes this task achievable. To assess the endurance squeeze pressure, measurements are taken during a 5-s- to 1-min-long squeeze. In health it is normal to be able to generate an increased pressure for at least 5 s. The fatigability of the external sphincter is qualitatively abnormal if a raised pressure is generated for less than 5 s.

Marcello et al. developed a quantitative measure of fatigability [50]. By calculating the rate of fatigue rate (using linear regression on the mean pressure over 1 s periods throughout the endurance squeeze), a measurement can be derived which represents the time it would take for the pressure to return to resting pressure. For reference the equation to derive the fatigue rate index (FRI) is:

$$\mathrm{FRI} = \left(\mathrm{P}_{\mathrm{MaxSqueeze}} - \mathrm{P}_{\mathrm{MaxResting}}\right) \times \left(\mathrm{Fatigue\,Rate}\right)^{-1}$$

As with maximum squeeze pressures, there are significant differences in FRI between incontinent and healthy volunteers, but there is still a large degree of overlap [51].

Involuntary Squeeze

Increased intra-anal pressure is observed during manoeuvres that increase intra-abdominal and therefore intrarectal pressure in order to maintain a positive anorectal pressure gradient. Commonly used manoeuvres are the cough and the Valsalva manoeuvre. Intra-anal pressure increase during a cough is normally above around 50 cmH_2O.

This measures an important role of the external anal sphincter in the maintenance of continence. The pressure within the rectum is not constant and rises considerably during episodes of raised intra-abdominal pressure and during rectal filling. The rise in the rectal pressure has to be responded to in order to prevent incontinent episodes. This led early theorists to postulate the existence of some sort of valve mechanism which enables the pressure within the rectum to exceed that of the anal canal without incontinence. The 'flap valve' was born from the concept of the anorectal angle that is generated by pelvic floor muscles, specifically puborectalis. It was postulated that the anterior wall of the rectum is forced down on the proximal anal canal to seal the anal canal preventing leakage of rectal contents. However, the external anal sphincter has been shown to contract in response to increased abdominal pressure maintaining a positive anorectal pressure gradient [52]. If a positive anorectal pressure gradient is not maintained, there is leakage of stools. Imaging the rectum and anus during the Valsalva manoeuvre confirms that the anterior rectal wall does not come into contact with the upper anal canal [53]. All of this makes the 'flap theory' an unlikely mechanism in the maintenance of continence during raises in the intra-abdominal pressure. The role of puborectalis and the anorectal angle is, however, central to the continence mechanism. Incontinence consistently results from division of puborectalis. The contraction of the external anal sphincter in response to raised intra-abdominal pressure is likely to be a learned response rather than a spinal or intramural reflex.

Straining/Defaecatory Manoeuvres

Balloon expulsion and anal canal pressure measurements during straining are relatively easy investigations to perform on patients with evacuatory difficulty. In order to understand the limitations of these tests, the sequence of visceral and somatic afferent and efferent responses involved in normal defaecation needs to be appreciated.

Normal defaecation is initiated with the sensation of urge or fullness following delivery of colonic contents into the rectum. The act of defaecation can be deferred with voluntary contraction of the pelvic floor and the external anal sphincter. Deferral can cause proximal movement of the stool back into the sigmoid colon or just allows time for the rectal smooth muscle to undergo receptive relaxation reducing rectal pressure.

Once the decision has been made to evacuate the rectum, the process of reversing the normally positive anorectal pressure gradient has to take place. Behavioural mechanisms such as sitting allow the anorectal angle to widen facilitating the passage of solid stool. Contraction of the abdominal wall muscles raises the intra-abdominal pressure which is transmitted to the rectum which deforms the anterior wall making it concave. The anal canal shortens in length, becomes funnel shaped and opens and with further increase in the intra-abdominal pressure allows the rectum to empty fully. There are various visceral elements to the defaecatory mechanism. The rectum has large motor complexes causing contraction, and the sigmoid and left colon have been shown to empty by an average of 32 % [54].

Defaecation is therefore dependent on the integration of anorectal sensation and visceral motor activity of the colon and rectum in addition to the somatic control of the abdominal wall and pelvic floor.

Our knowledge of normal defaecation is based largely on evacuation proctography, and the limitations of this investigation reflect the limitations in the understanding of normal defaecation. There is a significant psychological aspect in the investigation of defaecation with the inevitable embarrassment of being observed whilst voiding. In addition, the normal urge to defaecate is often absent, as are the physiological changes of the rectum and colon that accompany defaecation. Thus all laboratory investigations in defaecation assess the voluntary aspect of defaecation in the absence of any upstream colonic activity.

Anatomical abnormalities such as rectoceles and rectal intussusception are a frequent finding in proctography in patients with an evacuatory problem. Understanding of these anatomical pathologies is essential to correctly interpret physiological measures of evacuation such as balloon expulsion.

A rectocele is a bulge in the rectovaginal septum during defaecation. Evacuation proctography in normal individuals has shown up to 81 % of women have rectoceles with 48 % being greater than 1 cm in depth and 5 % being greater than 2 cm in depth [55, 56]. The size of the rectocele is a function of the stiffness of the rectovaginal septum and the maximum rectovaginal pressure gradient generated during straining. If there is significant external anal sphincter contraction during attempted defaecation (anismus), then rectal pressure will be allowed to increase further than in someone who is able to fully relax their anal canal. Rectocele size is therefore not an absolute figure but represents the distance that a tissue moves in response to a force. However, it is generally considered that a rectocele of less than 2 cm is unlikely to cause symptoms and that a rectocele of greater than 3.5 cm is large. The functional significance of a rectocele is probably related to size; however, the concept of 'trapping' has been introduced to further ascertain its functional significance. Trapping is defined as retention of greater than 10 % of the contents of the rectocele following evacuation of the rectal contents. The contents of the rectocele normally disappear following defaecation in isolation in a normal toilet, questioning the utility of the concept of trapping [57].

Although the size and extent of trapping are useful indicators for the functional significance of a rectocele, they are no substitute for a good history including feeling of a bulge in the vagina and eliciting a history of vaginal digitation to assist defaecation, the latter providing compelling evidence for a functionally significant rectocele [58].

Rectal intussusception can be graded depending on whether it is circumferential, the length of the infolding and the position of the leading edge being either intrarectal or intra-anal. Greater than 50 % of normal subjects have a grade 4 intussusception, defined as a greater-than-3 mm circumferential intrarectal intussusception [56]. This implies that a degree of intussusception is part of normal defaecation. Intra-anal intussusception is thought to be pathological and probably forms a continuum with rectal prolapse. Chronic intra-anal intussusception and rectal prolapse are often associated with thickening of the internal anal sphincter on endoanal ultrasound [59].

Rectal prolapse is idiopathic in adults although chronic excessive straining, eating disorders, connective tissue disorders and joint hypermobility

syndromes are all risk factors. Clinical features include a sensation of a lump on staining, either spontaneously reducing or requiring digital reduction. Rectal prolapse should always be excluded in elderly patients with passive incontinence especially in the context of chronic constipation. Significant proportion are missed on clinical examination, and it is important that the patient is examined on a toilet or commode and the patient is given enough time alone to strain to the same extent they would normally do in order to defaecate.

Solitary rectal ulcer syndrome is defined by the presence of a rectal ulcer up to 12 cm from the anal verge, and histologically it is consistent with ischaemia or trauma [60].

The pathophysiology of solitary rectal ulcer syndrome is unclear but may be linked to rectal intussusception, with anal digitation possibly being a contributory factor in some cases. Certainly a pathologically thickened internal sphincter on endoanal ultrasound is almost always present.

One of the initial physiological studies of patients with evacuatory difficulty was Preston et al.'s study. They included patients with slow-transit constipation and compared them to normal subjects using electromyography of puborectalis and balloon expulsion [61]. They found that puborectalis EMG activity decreased during straining in normal individuals but increased in patients with slow-transit constipation. They termed this anismus, likening the condition to spasm of the pelvic floor muscles in vaginismus. This finding has been replicated in other studies using EMG, manometry and evacuation proctography. Delayed initiation of evacuation and incomplete and prolonged evacuation are the most accurate indicators of anismus in evacuation proctography [62]. However, further studies have shown that the pelvic floor and the EAS do not relax in up to 50 % of individuals with normal defaecation [63]. The functional significance of absence of relaxation or contraction of puborectalis during defaecation is unknown. It may be that it is normal to have a degree of contraction during normal defaecation, or it may be that this phenomenon only occurs under experimental conditions. It has been shown using high-resolution anal manometry that patients with increased intra-anal pressures on straining whilst in the left lateral position can have decreased intra-anal pressure on straining whilst in the seated position [64]. There is also debate on whether puborectalis contraction is in response to the absence of stool passing through the anal canal rather than a cause of the constipation. Certainly deferring defaecation has been shown to slow colonic transit in normal individuals, establishing colonic transit and rectal evacuation as inextricably linked [65].

Despite the controversy surrounding the functional significance of anismus, it has firmly established itself as a diagnosis in patients with obstructive defaecation syndrome. It responds well to defaecatory retraining and behavioural techniques used in biofeedback therapy, and these aspects should be addressed prior to surgical correction of anatomical causes of obstructive defaecation.

Rectoanal Inhibitory Reflex (RAIR)

Rectal distension induces a relaxation response in the internal anal sphincter and is termed the rectoanal inhibitory reflex (RAIR) [66]. It is elicited by rapid insufflation and desufflation of 50 ml of air into a balloon positioned in the distal rectum during anal manometry at the level of the proximal high-pressure zone. A drop of at least 25 % of resting pressure has to occur with subsequent restoration to at least two thirds of resting pressure for the RAIR to be deemed present [67]. Smaller volumes of distension result in a transient reduction in anal canal pressures, and larger volumes produce a more sustained response.

In addition to the internal anal sphincter relaxation, there is a contraction of the external anal sphincter termed the inflation reflex or rectoanal excitatory response (RAER). However, this is not present in spinal injury patients where there is no EMG activity within the EAS during rectal distension [33]. The inflation reflex is less likely to occur during sleep and can be voluntarily omitted [68]. The contraction of the EAS during rectal distension is therefore a learned excitatory response rather than a spinal reflex.

The rectal mucosal receptors respond to the distension stimulus, and this is transmitted along

the intramural nerve plexus to initiate relaxation in the internal anal sphincter. Although the RAIR is mediated by the intramural plexus, it is modulated by the extrinsic autonomic supply, demonstrated by the lack of relationship between intensity of stimulus and anal canal relaxation in spinal injury patients.

The chemical mediators of the RAIR are controversial as with the tonic control of the IAS. It is likely that nitric oxide plays an important role although the RAIR is still present in NO synthase knockout mice indicating that it is not the exclusive NANC neurotransmitter responsible for the RAIR [40].

It has been proposed that IAS relaxations act as part of a sensory system allowing rectal contents to come in contact with the sensitive anal canal mucosa acting as a 'sampling reflex' [69]. This enables the contents of the rectum to be identified and flatus selectively expelled without soiling.

In clinical practice the RAIR is used in the diagnosis of Hirschsprung's. This can produce false positives (absence of RAIR without Hirschsprung's) which has led to investigating the benefits of other stimuli including free air, electrical and cold water, none of which have had widespread uptake. False positives (i.e. apparent failure of relaxation of the sphincter) can occur in patient with very low resting pressures. It is the pressure not the volume within the rectal balloon which has to exceed a threshold figure to elicit the inhibitory response. Therefore volumes have to be higher in patients with a megarectum to avoid false positives in this group.

Quantitative analysis of the RAIR (providing the percentage decrease in pressure, duration and volume needed to elicit) has been used in the study of patients with faecal incontinence with normal sphincter structure, so-called idiopathic faecal incontinence [67]. Studies have shown abnormalities in IAS function in this group, although many of them are contradictory. Several studies have shown IAS relaxation occurs with reduced rectal stimuli and with a greater degree of relaxation in patients with idiopathic faecal incontinence compared to normal subjects [70]. This is contrasted with the findings that the 'sampling reflex' occurs less frequently in patients with faecal incontinence [71] and the RAIR is often absent in faecal incontinence especially in patients with very low resting pressures. Other findings of interest include the delay between IAS relaxation and the sensation of rectal fullness or urge.

Reflex activity dysfunction has been noted in pruritus ani with increased depth and duration of IAS relaxation in response to rectal distension [72]. Symptoms start within an hour of an exaggerated IAS relaxation suggesting that this allows the passive leakage of irritative faecal matter.

There is also conflicting evidence on the role of the RAIR and 'sampling reflex' in the ability to discriminate rectal contents. Some studies have found no effect of the loss of RAIR after rectal anastomosis on discriminatory ability, but others have found that when the RAIR returns after the intramural plexus bridges the anastomosis, the discriminatory ability increases [73, 74]. The conflicting results of anorectal physiology can probably be explained by heterogeneous study methodology, imprecise measurements and large variation in health and disease.

Advanced Manometric Techniques

Vector Volume Manometry

Vector manometry involves withdrawing (commonly using a mechanical puller) a radially arranged eight-channel anorectal manometry catheter through the length of the anal canal. The resulting data can be used to assess sphincter symmetry in an attempt to differentiate sphincter defects from atrophy and to generate an addition quantitative measure called the vector volume. The distance between pressure measurements is dictated to by the speed of the mechanical puller (3–25 mm/s) and the sampling frequency of pressure measurements (20–100 Hz) resulting in a pressure measurement taken every 0.03–1.25 mm depending on the combination of puller speed and sampling frequency. Mechanical puller withdrawal at 25 mm/s has been shown to provide the optimum precision for vector volume manometry [75].

Radial asymmetry index (RAI) is a quantitative measure of the radial symmetry and can be calculated at any level in the anal canal but most commonly refers to the level at which the highest resting pressure is generated. It is expressed as a percentage with 0 % indicating no asymmetry, with all channels recording the same value. The principle is that an asymmetrical sphincter is more likely to have a sphincter defect although this is highly inaccurate compared to the gold standard of endoanal ultrasound. For reference the equation to calculate this is as follows [75]:

$$\text{Radial asymmetry index} = 100 - \left(\frac{(P_1 + P_2 + P_3 + \cdots P_n)}{(P_{\max} \times n) \times 100} \right)$$

where n = Number of radially arranged pressure sensors $P_{\max}$ = Maximum resting pressure at the level of interest

The vector volume is the volume of the 3D shape generated and provides a value which reflects the overall length and symmetry of the sphincter and the length of the HPZ in addition to the maximum pressure. The cross-sectional area at each level is calculated by addition of the individual segments, for an 8-channel catheter [76]:

$$\text{Radial area} = 0.5 \times \sin(45) \times \left(\frac{(P_1 \times P_2) + (P_2 \times P_3) \cdots}{(P_7 + P_8) + (P_8 + P_1)} \right)$$

The volume is then calculated by summing cross-sectional areas from every level and multiplying by the distance between each level.

High-Resolution Manometry

High-resolution manometry uses a catheter with a large number of pressure sensors spaced less than 0.5 mm apart along the length of the catheter. This enables the pressure between the data points to be accurately interpolated which allows complete definition of the intra-anal pressure environment. The resulting data is displayed on a topographical plot (Clouse plot) to allow easier pattern recognition. It is a three-dimensional measurement with the variables of pressure (displayed as the colour), distance into the anal canal (y-axis) and time (x-axis).

Both water-perfused and solid-state catheters can be used. Solid-state high-resolution catheters can employ over 100 micro-pressure transducers. Water-perfused systems generally have a limit of 20 channels, but the catheters are disposable and cheaper.

High-resolution manometry was developed initially for oesophageal manometry. Advanced quantitative measures were developed which enabled existing diseases to be subclassified [77, 78]. Jones et al. compared high-resolution anal manometry with conventional water-perfused manometry and found that measurements correlated. It has yet to be accepted as a standard clinical tool [79].

Sensory Measurements

Normal anorectal sensation is essential for continence and defaecatory mechanisms to work effectively.

Assessment of Rectal Sensation to Distension

Rectal sensation to distension is most commonly assessed by inflating an intrarectal balloon. The volume which elicits the first sensation of balloon expansion (threshold), urge and discomfort can then be recorded. The pressure required to elicit these sensations can also be measured using a barostat and may be more precise. When using a barostat for sensory measurements, it is recommended that random phasic distensions are used with 60 s of inflation at a random pressure followed by a wait of 60 s before starting another random distension [80]. A graduated scale should be used to report symptoms during the distension. Random phasic distension adds unpredictability which aims to reduce anticipation bias. It is however less physiological as the rectum normally fills progressively rather than phasically. The barostat measures the volume and the pressure required to elicit threshold, urge and maximum tolerated volumes; however it does not measure

the dimensions of the rectum at which these sensations occur. Impedance planimetry measures the cross-sectional area of an intrarectal bag during different pressure distensions. Using this technique which is not part of routine clinical practice has shown that rectal sensation is more closely linked to the circumferential rectal strain than rectal volume or pressure. This suggests that rectal distension sensation is mediated via length-dependent mechanical receptors [81].

Anorectal Sensation to Electrical Stimulation

Both anal and rectal sensation can be assessed with mucosal electrical stimulation [82]. This is performed using a probe with two electrodes between which a small electrical potential is applied generating an alternating square wave with a 5–20 Hz frequency. The potential across the electrodes is increased to generate up to 20 mA for anal sensation and 50 mA for rectal sensation. The amplitude is gradually increased to find the lowest current required to elicit the stimulus. The sensation elicited on applying this electrical stimulation to the anal canal is often described as a tingling, tapping or buzzing sensation. This differs from rectal stimulation which elicits an ache in the lower abdomen or back.

Anorectal sensory measurements are generally more precise than the measurements of the anorectal motor function such as manometry [83–85].

Physiology of Rectal Sensation

In contrast to the anal canal, Duthie and Gairns when studying the rectum found no free nerve endings and few or no organised nerve endings resulting in a poor sensitivity to pain, touch and temperature. Goligher et al. described the ability to detect balloon distension in the colon and rectum [86]. Distension in the colon results in a sensation of discomfort in the lower abdomen in contrast to distension of the rectum which results in a sensation of flatus or the desire to defaecate. The closer to the anal canal the balloon was placed, the lower the pressure was needed to elicit these sensations. The origin of these sensations in response to distension in the absence of nerve endings within the rectum is still unknown. One explanation is that the rectal stretching causes stretching of the pelvic floor. Walls described 'stretch receptors' in levator ani and the external anal sphincter [87]. This could explain the ability of patients with an ileopouch anal anastomosis to detect balloon distension and why this distension elicits the same sensation of fullness and urgency.

Rectal distension is transmitted via the parasympathetic nervous system to the S 2, 3 and 4 nerve roots.

Function of Normal Anorectal Sensation

Several times an hour, there is equalisation of the anorectal pressure gradient during which the functional anal canal shortens allowing rectal contents to come into contact with the sensitive anal mucosa. This is termed the 'sampling reflex' [69]. It is hypothesised that this allows discrimination between flatus, liquid stools and solid stools. The ability of the anal canal to differentiate minor changes in temperature is one modality through which this could be achieved. Elsewhere in the body the appreciation of 'wetness' and the distinction between gas, liquid and solid are reliant on temperature sensation. As flatus, stools with a high water content and solid stools all have different thermal conduction, it is possible that temperature is the modality used in detecting rectal contents. The importance of anal sensation in continence is questioned by a study by Read et al. who abolished anal sensation by applying topical local anaesthetic gel [88]. The ability of normal volunteers to retain a saline enema was not hindered; in fact it improved in two cases.

Pathophysiology of Anorectal Sensation

Baldi et al. and Kamm et al. have shown reduced rectal sensation as tested by balloon and electrical stimulation in some patients with idiopathic constipation [89, 90]. This suggests a sensory neuropathy. This could be within the intrinsic supply within the rectal wall or the extrinsic nerve supply. The myenteric plexus in these individuals is abnormal although this may be secondary to abnormalities in the extrinsic supply.

Pudendal neuropathy in incontinence is an established phenomenon in patients with idiopathic faecal incontinence. As the pudendal nerve is mixed and contains the afferent fibres from the anal canal, it follows that the anal canal sensation should be impaired in these patients. Indeed this has been shown, with reduced mucosal electrosensitivity and reduced temperature sensibility.

The role of rectal hypersensitivity in faecal urge incontinence and urgency has been investigated. Chan et al. have found a small subset of individuals with urge faecal incontinence who have enhanced rectal perception of rectal distension which worsens stool frequency and urgency [91]. They went on to find that patients with rectal hypersensitivity and faecal urgency have increased number of polymodal sensory fibres expressing TRPV1 expressed on afferent C fibres [92]. This may represent an abnormal response to nerve damage.

Advanced Physiological Measurements

These advanced physiological measurements provide further insight into the function of supra-sphincteric mechanisms of continence such as rectal compliance and pudendal neuropathy with neurophysiological measurements. They do not form part of routine clinical measurements but are frequently used in research in the understanding of pathophysiology and mechanisms of action of therapeutic interventions.

Rectal Compliance

The rectum's role in the storage of faeces prior to defaecation is central to the normal functioning of the continence and defaecatory mechanisms. The reservoir function is characterised by the small rise in pressure with increasing volume. Compliance is the term that describes the relationship between pressure and volume. It is defined as the ability of an organ to stretch to an imposed force.

The rectum has several factors which influence its ability to stretch. When distending the rectum, the connective tissues, the smooth muscle and the extra-rectal tissue all stretch and contribute to the overall rectal compliance.

Rectal smooth muscle is tonically active and can respond to distension with relaxation (receptive relaxation) and with contraction (rectal reactivity). The ability of the rectal smooth muscle to stretch is therefore time dependent. The ability of the rectal connective tissue to stretch varies with distension. As a result calculating the compliance of the rectum can be a complicated process. It is complicated further by the concept of circumferential wall tension. This is generally considered to be the biomechanical property which best represents tone. It is the radius of the organ multiplied by the transmural pressure, in this case the abdominal pressure subtracted from the rectal pressure. There is no mathematical relationship between compliance and circumferential wall tension, although they are conceptually related.

Barostat Measurement of Rectal Compliance

The barostat machine has a pump which can rapidly inject or withdraw air from a balloon in order to maintain an intraballoon pressure within a tight pressure window. The pressure is measured within the balloon via a separate line. The volume within the balloon is then constantly recorded.

There are many differences in the equipment and distension protocols in barostat measurements. The rectal balloon can be either an elastic balloon or an inelastic bag. An elastic balloon has been widely used in the past. It has rapid increase in pressure for small volumes followed by a small increase in pressure with further inflation. It therefore has a predictable but significant influence on compliance. An inelastic bag has an 'infinite compliance' below the capacity of the bag and a rapid rise in pressure at its capacity. It is therefore important to use a bag that functions well below its capacity. It is generally recommended when used in the rectum that a spherical oversized inelastic bag is used in preference to an elastic balloon [80, 93]. This is because pressures

used in barostat testing normally fall into the elastic range of the balloon. In addition elastic balloons extend longitudinally to a greater extent which falsely lowers compliance as the added volume does not stretch the rectum but extends the length of the balloon. For this reason bags are fixed to the rigid catheter at each end to prevent longitudinal extension. The length of the bag has a major influence on the compliance as it affects the amount of rectum that is being stretched. This must be kept constant if comparisons are being made within or between individuals.

The pressure which distends the bag sufficiently so that it is in contact with the rectum circumferentially is called the minimal operating pressure or the standard operating pressure. This can be set figure, normally 6–10 mmHg. It can be individualised by slowly increasing the intra-bag pressure until a change in the volume of the bag can be detected consistent with the abdominal pressure changes associated with respiration. Inflating the bag within the rectum prior to the recording inflation protocol, known as conditioning, has been shown to improve the precision of compliance testing [94].

The pressure is set using a computer interface. The pressure can increase constantly above the standard operating pressure up to 40 mmHg. This is a ramp distension named after the shape of the pressure trace. The alternative is to have a stepwise increase in pressure with 30 s to a minute at each pressure level increasing typically at 4 mmHg at each step. The volume can be averaged over the whole pressure step or during the last 5–10 s. The third option is to increase the pressure in a phasic manner reducing the pressure to zero in between distensions.

The barostat can be used to ascertain sensory thresholds. In this case random phasic distensions have advantages over cumulative ramp or stepwise distensions as it reduces response bias with anticipating higher intensity of stimulation. It does not reflect the normal filling of the rectum and is therefore less physiological.

The difference between ramp and stepwise is the length of time at each pressure level. This in turn affects the response of the rectal smooth muscle. During stepwise inflation the rectal smooth muscle undergoes receptive relaxation accommodating further volume at a given pressure. To eliminate receptive relaxation, the high rates of ramp inflation have to be used. High rates of rectal inflation however can cause rectal smooth muscle contraction lasting up to 20 s. In addition it may be preferable to ascertain the degree of rectal smooth muscle relaxation. Ramp and stepwise inflation protocols therefore calculate the rectal compliance with the smooth muscle in different states of tonic activity. It is therefore important that the inflation protocol is kept constant if valid comparisons can be made within and between individuals.

Rectal Compliance Curve

The rectum consistently has a sigmoidal or triphasic compliance curve. The initial steep rise in the pressure with volume is thought to represent the elastic properties of the contracted rectal wall. The subsequent plateau is a result of receptive relaxation of the rectum. This is where the rectal smooth muscle reacts to the increase in wall tension by relaxing increasing the rectal compliance. The final steep rise in pressure occurs when the limits of smooth muscle relaxation have been reached and represents the elastic properties of the fully relaxed rectal wall.

Compliance describes the relationship between pressure and volume. As described above the relationship is not linear which makes defining compliance by a single number difficult. One method is to describe the gradient at specific point. This is usually during the plateau phase and represents the greatest compliance during the distension sequence. Another option is to logarithmically transform the data to create a linear relationship allowing a simple equation to describe the compliance curve.

Reproducibility of Barostat Measurements

Several studies have shown that barostat measurements are relatively reproducible on a day-to-day basis within the same institution. Reproducibility improves by using an oversize noncompliant bag rather than an elastic balloon. There is, however, a very large interindividual and inter-institutional

variation in healthy controls. This could be a result of the differing methodologies and the effect of differing rectal capacity has on compliance. It has been suggested that the compliance should be standardised with rectal capacity to help reduce this variation [93].

Physiological Factors Influencing Rectal Compliance

Although the rectal compliance curve has a consistent shape, the curve can move up or down within individuals causing alteration in the rectal compliance. In a study using a ramp distension protocol and an elastic balloon found that the rate of balloon filling influences rectal compliance [95]. Lower rates of rectal filling are associated with a higher rectal compliance. This is possibly due to the time-dependent relaxation of the rectal smooth muscle being too slow to respond maximally at higher rectal filling rates. This rate-dependent change in compliance, however, was not replicated by Fox et al. using a mechanical barostat [93]. Bell et al. found rectal compliance decreased following meals and increased with glucagon [96]. They also found that administering neostigmine a parasympathomimetic reduced rectal compliance, the mechanism for which is via rectal smooth muscle contraction.

Pathophysiology of Rectal Compliance

Rectal compliance is possibly reduced in diarrhoea-predominant IBS. Rao et al. demonstrated that rectal compliance is reduced in ulcerative colitis in addition to an increased rectal contraction in response to distension [96, 97]. Following induction of remission, the rectal compliance and rectal reactivity returned to normal indicating that the inflammatory process affects the tonic activity of the rectal smooth muscle causing a reversible reduction in rectal compliance. Pelvic irradiation has also been shown to reduce rectal compliance [98].

The postprandial increase in rectal tone is lower in chronically constipated patients. Although there are disease conditions which affect rectal compliance, firm conclusions are difficult to draw as there are no accepted indications for the routine clinical use of the barostat.

Impedance Planimetry

Rectal impedance planimetry studies calculate the diameter or cross-sectional area of an intrarectal bag during a distension sequence. Volume measurements in spherical organs such as the bladder have a direct relationship with radius. Volume measurements in tubular organs such as the rectum may not be related to the radius because the shape may expand longitudinally. The volume can therefore increase by lengthening whilst having a constant radius. In addition the compressibility of air means that even in spherical distensions, there may not be a direct relationship between the volume infiltrated and radius. The relationship between volume and radius is therefore unknown in barostat measurements. Impedance planimetry measures the cross-sectional area which enables the circumferential wall tension to be calculated. This may be a more physiologically meaningful representation of the biomechanical properties of the rectum.

Impedance planimetry has demonstrated a linear relationship between wall tension and distension pressure [99]. In addition impedance planimetry studies have shown the compliance of the rectum decreases in a non-linear fashion with distension pressure, which differs from that obtained with barostat measurements. This raises questions about the validity of the traditional sigmoidal-shaped compliance curve of the rectum obtained using the barostat. It is unknown whether this represents the true biomechanical properties of the rectum or results from the complexities of non-uniform relationship between volume and radius.

Impedance planimetry has been used in studies investigating rectal sensation. They have shown that sensory thresholds are likely to be mediated by length-dependent mechanical nociceptors rather than pressure- or tension-mediated mechanism [81].

Neurophysiology

EMG

Single-fibre EMG calculating EAS fibre density is another method of assessing denervation and

reinnervation of the EAS. It is used almost exclusively as a research tool and has not made it into routine clinical practice, maybe because of the invasive nature and complexity. Conventional EMG can be used to quantify the reinnervation of the EAS by detecting prolongation in the duration of the motor unit potential.

Electromyography (EMG) is not routinely performed to assess the IAS, although it is a useful research tool in distinguishing IAS from EAS activity.

PNTMLs

Pudendal nerve terminal motor latencies (PNTMLs) are the most commonly performed measure of pudendal neuropathy. The technique was first described by Kiff and Swash [100]. The pudendal nerve is stimulated as it passes over the ischial spine using a specially designed St. Marks electrode attached to the index finger of the assessor. In addition to the stimulating electrode, the St. Marks electrode has a surface EMG electrode which sits on the base of the assessor's index finger and measures external sphincter activity. The delay between the electrical stimulation of the pudendal nerve and the EMG activity of the external anal sphincter is the pudendal nerve terminal motor latency. The test is rarely used routinely as it does not reliably reflect the pudendal nerve damage. This may be because PNTMLs measure the speed of nerve conduction which involves the fastest nerve fibres which are least susceptible to damage. The test is also very operator dependent, with poor reproducibility.

Conclusions

Anorectal physiological measurements provide a useful quantitative measure of a specific anorectal function. Continence and defaecation however require the correct functioning and co-ordination of a multitude of motor and sensory sphincteric and suprasphincteric apparatus. It is only in the context of the patient's symptoms, thorough examination and radiological investigations that physiological measurements can be correctly interpreted.

References

1. Stewart WF, Liberman JN, Sandler RS, Woods MS, Stemhagen A, Chee E, et al. Epidemiology of constipation (EPOC) study in the United States: relation of clinical subtypes to sociodemographic features. Am J Gastroenterol. 1999;94(12):3530–40.
2. Longstreth GF, Thompson WG, Chey WD, Houghton LA, Mearin F, Spiller RC. Functional bowel disorders. Gastroenterology. 2006;130(5):1480–91.
3. Lubowski DZ, King DW. Obstructed defecation: current status of pathophysiology and management. Aust N Z J Surg. 1995;65(2):87–92.
4. Gladman MA, Lunniss PJ, Scott SM, Swash M. Rectal hyposensitivity. Am J Gastroenterol. 2006;101(5):1140–51.
5. Rao SS. Dyssynergic defecation. Gastroenterol Clin North Am. 2001;30(1):97–114.
6. Altomare D, Portincasa P, Rinaldi M, Ciaula A, Martinelli E, Amoruso A, et al. Slow-transit constipation. Dis Colon Rectum. 1999;42(2):231–40.
7. Agachan F, Chen T, Pfeifer J, Reissman P, Wexner SD. A constipation scoring system to simplify evaluation and management of constipated patients. Dis Colon Rectum. 1996;39(6):681–5.
8. Varma M, Wang J, Berian J, Patterson T, McCrea G, Hart S. The constipation severity instrument: a validated measure. Dis Colon Rectum. 2008;51(2):162–72.
9. Nelson R, Norton N, Cautley E, Furner S. Community-based prevalence of anal incontinence. JAMA. 1995; 274(7):559–61.
10. Roberts RO, Jacobsen SJ, Reilly WT, Pemberton JH, Lieber MM, Talley NJ. Prevalence of combined fecal and urinary incontinence: a community-based study. J Am Geriatr Soc. 1999;47(7):837–41.
11. Grover M, Busby-Whitehead J, Palmer Rn MH, Heymen S, Palsson OS, Goode PS, et al. Survey of geriatricians on the effect of fecal incontinence on nursing home referral. J Am Geriatr Soc. 2010;58(6): 1058–62.
12. Kamm MA. Obstetric damage and faecal incontinence. Lancet. 1994;344(8924):730–3.
13. Macarthur C, Glazener C, Lancashire R, Herbison P, Wilson D, Grant A. Faecal incontinence and mode of first and subsequent delivery: a six-year longitudinal study. BJOG. 2005;112(8):1075–82.
14. Sultan AH, Kamm MA, Hudson CN, Bartram CI. Third degree obstetric anal sphincter tears: risk factors and outcome of primary repair. BMJ. 1994;308(6933):887–91.
15. Cheng YW, Hopkins LM, Caughey AB. How long is too long: does a prolonged second stage of labor in nulliparous women affect maternal and neonatal outcomes? Am J Obstet Gynecol. 2004;191(3):933–8.
16. Elfaghi I, Johansson-Ernste B, Rydhstroem H. Rupture of the sphincter ani: the recurrence rate in second delivery. BJOG. 2004;111(12):1361–4.
17. de Parades V, Etienney I, Thabut D, Beaulieu S, Tawk M, Assemekang B, et al. Anal sphincter injury after

forceps delivery: myth or reality? A prospective ultrasound study of 93 females. Dis Colon Rectum. 2004;47(1):24–34.
18. Sultan AH, Kamm MA, Bartram CI, Hudson CN. Anal sphincter trauma during instrumental delivery. Int J Gynaecol Obstet. 1993;43(3):263–70.
19. Signorello LB, Harlow BL, Chekos AK, Repke JT. Midline episiotomy and anal incontinence: retrospective cohort study. BMJ. 2000;320(7227):86–90.
20. Nelson RL, Furner SE, Westercamp M, Farquhar C. Cesarean delivery for the prevention of anal incontinence. Cochrane Database Syst Rev. 2010;(2): CD006756.
21. McKenna DS, Ester JB, Fischer JR. Elective cesarean delivery for women with a previous anal sphincter rupture. Am J Obstet Gynecol. 2003;189(5): 1251–6.
22. Pescatori M, Anastasio G, Bottini C, Mentasti A. New grading and scoring for anal incontinence. Evaluation of 335 patients. Dis Colon Rectum. 1992;35(5): 482–7.
23. Jorge JM, Wexner SD. Etiology and management of fecal incontinence. Dis Colon Rectum. 1993;36(1):77–97.
24. Vaizey CJ, Carapeti E, Cahill JA, Kamm MA. Prospective comparison of faecal incontinence grading systems. Gut. 1999;44(1):77–80.
25. Dobben A, Terra M, Deutekom M, Gerhards M, Bijnen A, Felt-Bersma R, et al. Anal inspection and digital rectal examination compared to anorectal physiology tests and endoanal ultrasonography in evaluating fecal incontinence. Int J Colorectal Dis. 2007;22(7):783–90.
26. Siproudhis L, Ropert A, Vilotte J, Bretagne JF, Heresbach D, Raoul JL, et al. How accurate is clinical examination in diagnosing and quantifying pelvirectal disorders? A prospective study in a group of 50 patients complaining of defecatory difficulties. Dis Colon Rectum. 1993;36(5):430–8.
27. Tantiphlachiva K, Rao P, Attaluri A, Rao SSC. Digital rectal examination is a useful tool for identifying patients with dyssynergia. Clin Gastroenterol Hepatol. 2010;8(11):955–60.
28. Minguez M, Herreros B, Sanchiz V, Hernandez V, Almela P, Anon R, et al. Predictive value of the balloon expulsion test for excluding the diagnosis of pelvic floor dyssynergia in constipation. Gastroenterology. 2004;126(1):57–62.
29. Bennett RC, Duthie HL. The functional importance of the internal anal sphincter. Br J Surg. 1964;51: 355–7.
30. Duthie HL, Watts JM. Contribution of the external anal sphincter to the pressure zone in the anal canal. Gut. 1965;6:64–8.
31. Frenckner B, Euler CV. Influence of pudendal block on the function of the anal sphincters. Gut. 1975; 16(6):482–9.
32. Floyd WF, Walls EW. Electromyography of the sphincter ani externus in man. J Physiol. 1953;122(3): 599–609.
33. Weber J, Beuret-Blanquart F, Ducrotte P, Touchais JY, Denis P. External anal sphincter function in spinal patients. Electromyographic and manometric study. Dis Colon Rectum. 1991;34(5):409–15.
34. Lestar B, Penninckx F, Rigauts H, Kerremans R. The internal anal sphincter can not close the anal canal completely. Int J Colorectal Dis. 1992;7(3): 159–61.
35. Enck P, Eggers E, Koletzko S, Erckenbrecht JF. Spontaneous variation of anal "resting" pressure in healthy humans. Am J Physiol. 1991;261(5 Pt 1): G823–6.
36. Lubowski DZ, Nicholls RJ, Swash M, Jordan MJ. Neural control of internal anal sphincter function. Br J Surg. 1987;74(8):668–70.
37. Parks AG, Fishlock DJ, Cameron JD, May H. Preliminary investigation of the pharmacology of the human internal anal sphincter. Gut. 1969;10(8): 674–7.
38. Frenckner B, Ihre T. Influence of autonomic nerves on the internal and sphincter in man. Gut. 1976;17(4):306–12.
39. Speakman CT, Hoyle CH, Kamm MA, Henry MM, Nicholls RJ, Burnstock G. Neuropeptides in the internal anal sphincter in neurogenic faecal incontinence. Int J Colorectal Dis. 1993;8(4):201–5.
40. Jones OM, Brading AF, Mortensen NJ. Role of nitric oxide in anorectal function of normal and neuronal nitric oxide synthase knockout mice: a novel approach to anorectal disease. Dis Colon Rectum. 2003;46(7):963–70.
41. Rattan S, Culver PJ. Influence of loperamide on the internal anal sphincter in the opossum. Gastroenterology. 1987;93(1):121–8.
42. Holdsworth PJ, Sagar PM, Lewis WG, Williamson M, Johnston D. Internal anal sphincter activity after restorative proctocolectomy for ulcerative colitis: a study using continuous ambulatory manometry. Dis Colon Rectum. 1994;37(1):32–6.
43. Schouten WR, Briel JW, Auwerda JJ. Relationship between anal pressure and anodermal blood flow. The vascular pathogenesis of anal fissures. Dis Colon Rectum. 1994;37(7):664–9.
44. Lund JN, Scholefield JH. A randomised, prospective, double-blind, placebo-controlled trial of glyceryl trinitrate ointment in treatment of anal fissure. Lancet. 1997;349(9044):11–4.
45. McNamara MJ, Percy JP, Fielding IR. A manometric study of anal fissure treated by subcutaneous lateral internal sphincterotomy. Ann Surg. 1990;211(2): 235–8.
46. Bielecki K, Kolodziejczak M. A prospective randomized trial of diltiazem and glyceryltrinitrate ointment in the treatment of chronic anal fissure. Colorectal Dis. 2003;5(3):256–7.
47. Parks AG, Swash M, Urich H. Sphincter denervation in anorectal incontinence and rectal prolapse. Gut. 1977;18(8):656–65.
48. Snooks SJ, Henry MM, Swash M. Anorectal incontinence and rectal prolapse: differential assessment of

the innervation to puborectalis and external anal sphincter muscles. Gut. 1985;26(5):470–6.
49. Snooks SJ, Henry MM, Swash M. Faecal incontinence due to external anal sphincter division in childbirth is associated with damage to the innervation of the pelvic floor musculature: a double pathology. Br J Obstet Gynaecol. 1985;92(8):824–8.
50. Marcello P, Barrett R, Coller J, Schoetz D, Roberts P, Murray J, et al. Fatigue rate index as a new measurement of external sphincter function. Dis Colon Rectum. 1998;41(3):336–43.
51. Telford KJ, Ali ASM, Lymer K, Hosker GL, Kiff ES, Hill J. Fatigability of the external anal sphincter in anal incontinence. Dis Colon Rectum. 2004; 47(5):746–52.
52. Bannister JJ, Gibbons C, Read NW. Preservation of faecal continence during rises in intra-abdominal pressure: is there a role for the flap valve? Gut. 1987;28(10):1242–5.
53. Bartolo DC, Roe AM, Locke-Edmunds JC, Virjee J, Mortensen NJ. Flap-valve theory of anorectal continence. Br J Surg. 1986;73(12):1012–4.
54. Lubowski DZ, Meagher AP, Smart RC, Butler SP. Scintigraphic assessment of colonic function during defaecation. Int J Colorectal Dis. 1995;10(2):91–3.
55. Freimanis MG, Wald A, Caruana B, Bauman DH. Evacuation proctography in normal volunteers. Invest Radiol. 1991;26(6):581–5.
56. Shorvon PJ, McHugh S, Diamant NE, Somers S, Stevenson GW. Defecography in normal volunteers: results and implications. Gut. 1989;30(12):1737–49.
57. Greenberg T, Kelvin FM, Maglinte DD. Barium trapping in rectoceles: are we trapped by the wrong definition? Abdom Imaging. 2001;26(6):587–90.
58. Maglinte DD, Bartram C. Dynamic imaging of posterior compartment pelvic floor dysfunction by evacuation proctography: techniques, indications, results and limitations. Eur J Radiol. 2007;61(3):454–61.
59. Marshall M, Halligan S, Fotheringham T, Bartram C, Nicholls RJ. Predictive value of internal anal sphincter thickness for diagnosis of rectal intussusception in patients with solitary rectal ulcer syndrome. Br J Surg. 2002;89(10):1281–5.
60. Singh B, Mortensen NJM, Warren BF. Histopathological mimicry in mucosal prolapse. Histopathology. 2007;50(1):97–102.
61. Preston DM, Lennard-Jones JE. Anismus in chronic constipation. Dig Dis Sci. 1985;30(5):413–8.
62. Halligan S, Bartram CI, Park HJ, Kamm MA. Proctographic features of anismus. Radiology. 1995; 197(3):679–82.
63. Jones PN, Lubowski DZ, Swash M, Henry MM. Is paradoxical contraction of puborectalis muscle of functional importance? Dis Colon Rectum. 1987; 30(9):667–70.
64. Seo HI, Park JH, Sohn CI. Is abnormality in the conventional anorectal manometry really abnormal? J Neurogastroenterol Motil. 2010;16(2):213–4.
65. Klauser AG, Voderholzer WA, Heinrich CA, Schindlbeck NE, Muller-Lissner SA. Behavioral modification of colonic function. Can constipation be learned? Dig Dis Sci. 1990;35(10):1271–5.
66. Denny-Brown D, Robertson EG. 'An investigation of the nervous control of defecation' by Denny-Brown and Robertson: a classic paper revisited. 1935. Colorectal Dis. 2004;6(5):376–83.
67. Zbar A, Aslam M, Gold D, Gatzen C, Gosling A, Kmiot W. Parameters of the rectoanal inhibitory reflex in patients with idiopathic fecal incontinence and chronic constipation. Dis Colon Rectum. 1998;41(2):200–8.
68. Whitehead WE, Orr WC, Engel BT, Schuster MM. External anal sphincter response to rectal distention: learned response or reflex. Psychophysiology. 1982;19(1):57–62.
69. Duthie HL, Bennett RC. The relation of sensation in the anal canal to the functional anal sphincter: a possible factor in anal continence. Gut. 1963;4(2): 179–82.
70. Kaur G, Gardiner A, Duthie GS. Rectoanal reflex parameters in incontinence and constipation. Dis Colon Rectum. 2002;45(7):928–33.
71. Miller R, Bartolo DC, Cervero F, Mortensen NJ. Anorectal sampling: a comparison of normal and incontinent patients. Br J Surg. 1988;75(1):44–7.
72. Farouk R, Duthie GS, Pryde A, Bartolo DC. Abnormal transient internal sphincter relaxation in idiopathic pruritus ani: physiological evidence from ambulatory monitoring. Br J Surg. 1994;81(4): 603–6.
73. O'Riordain MG, Molloy RG, Gillen P, Horgan A, Kirwan WO. Rectoanal inhibitory reflex following low stapled anterior resection of the rectum. Dis Colon Rectum. 1992;35(9):874–8.
74. Saigusa N, Belin BM, Choi HJ, Gervaz P, Efron JE, Weiss EG, et al. Recovery of the rectoanal inhibitory reflex after restorative proctocolectomy: does it correlate with nocturnal continence? Dis Colon Rectum. 2003;46(2):168–72.
75. Schizas AMP, Emmanuel AV, Williams AB. Vector volume manometry – methods and normal values. Neurogastroenterol Motil. 2011;23(9):886–e393.
76. Perry RE, Blatchford GJ, Christensen MA, Thorson AG, Attwood SEA. Manometric diagnosis of anal sphincter injuries. Am J Surg. 1990;159(1):112–7.
77. Pandolfino JE, Ghosh SK, Zhang Q, Jarosz A, Shah N, Kahrilas PJ. Quantifying EGJ morphology and relaxation with high-resolution manometry: a study of 75 asymptomatic volunteers. Am J Physiol Gastrointest Liver Physiol. 2006;290(5):G1033–40.
78. Pandolfino JE, Fox MR, Bredenoord AJ, Kahrilas PJ. High-resolution manometry in clinical practice: utilizing pressure topography to classify oesophageal motility abnormalities. Neurogastroenterol Motil. 2009;21(8):796–806.
79. Jones MP, Post J, Crowell MD. High-resolution manometry in the evaluation of anorectal disorders: a simultaneous comparison with water-perfused manometry. Am J Gastroenterol. 2007;102(4): 850–5.

80. Whitehead WE, Delvaux M. Standardization of barostat procedures for testing smooth muscle tone and sensory thresholds in the gastrointestinal tract. The Working Team of Glaxo-Wellcome Research, UK. Dig Dis Sci. 1997;42(2):223–41.
81. Petersen P, Gao C, Arendt-Nielsen L, Gregersen H, Drewes AM. Pain intensity and biomechanical responses during ramp-controlled distension of the human rectum. Dig Dis Sci. 2003;48(7):1310–6.
82. Roe AM, Bartolo DC, Mortensen NJ. New method for assessment of anal sensation in various anorectal disorders. Br J Surg. 1986;73(4):310–2.
83. Bharucha AE, Seide B, Fox JC, Zinsmeister AR. Day-to-day reproducibility of anorectal sensorimotor assessments in healthy subjects. Neurogastroenterol Motil. 2004;16(2):241–50.
84. Diamant NE, Kamm MA, Wald A, Whitehead WE. AGA technical review on anorectal testing techniques. Gastroenterology. 1999;116(3):735–60.
85. Rogers J, Laurberg S, Misiewicz JJ, Henry MM, Swash M. Anorectal physiology validated: a repeatability study of the motor and sensory tests of anorectal function. Br J Surg. 1989;76(6):607–9.
86. Goligher JC, Hughes ES. Sensibility of the rectum and colon. Its role in the mechanism of anal continence. Lancet. 1951;1(6654):543–7.
87. Walls EW. Recent observations on the anatomy of the anal canal. Proc R Soc Med. 1959;52(Suppl): 85–7.
88. Read MG, Read NW. Role of anorectal sensation in preserving continence. Gut. 1982;23(4):345–7.
89. Kamm MA, Lennard-Jones JE. Rectal mucosal electrosensory testing – evidence for a rectal sensory neuropathy in idiopathic constipation. Dis Colon Rectum. 1990;33(5):419–23.
90. Baldi F, Ferrarini F, Corinaldesi R, Balestra R, Cassan M, Fenati GP, et al. Function of the internal anal sphincter and rectal sensitivity in idiopathic constipation. Digestion. 1982;24(1):14–22.
91. Chan CL, Scott SM, Williams NS, Lunniss PJ. Rectal hypersensitivity worsens stool frequency, urgency, and lifestyle in patients with urge fecal incontinence. Dis Colon Rectum. 2005;48(1): 134–40.
92. Chan CL, Facer P, Davis JB, Smith GD, Egerton J, Bountra C, et al. Sensory fibres expressing capsaicin receptor TRPV1 in patients with rectal hypersensitivity and faecal urgency. Lancet. 2003;361(9355):385–91.
93. Fox M, Thumshirn M, Fried M, Schwizer W. Barostat measurement of rectal compliance and capacity. Dis Colon Rectum. 2006;49(3):360–70.
94. Hammer HF, Phillips SF, Camilleri M, Hanson RB. Rectal tone, distensibility, and perception: reproducibility and response to different distensions. Am J Physiol. 1998;274(3 Pt 1):G584–90.
95. Sun WM, Read NW, Prior A, Daly JA, Cheah SK, Grundy D. Sensory and motor responses to rectal distension vary according to rate and pattern of balloon inflation. Gastroenterology. 1990;99(4): 1008–15.
96. Bell AM, Pemberton JH, Hanson RB, Zinsmeister AR. Variations in muscle tone of the human rectum: recordings with an electromechanical barostat. Am J Physiol. 1991;260(1 Pt 1):G17–25.
97. Suzuki H, Fujioka M. Rectal pressure and rectal compliance in ulcerative colitis. Jpn J Surg. 1982;12(1):79–81.
98. Varma JS, Smith AN, Busuttil A. Correlation of clinical and manometric abnormalities of rectal function following chronic radiation injury. Br J Surg. 1985;72(11):875–8.
99. Dall FH, Jorgensen CS, Houe D, Gregersen H, Djurhuus JC. Biomechanical wall properties of the human rectum. A study with impedance planimetry. Gut. 1993;34(11):1581–6.
100. Kiff ES, Swash M. Slowed conduction in the pudendal nerves in idiopathic (neurogenic) faecal incontinence. Br J Surg. 1984;71(8):614–6.

Imaging the Normal Anus

3

Michael J. Steward, Steve Halligan,
and Stuart A. Taylor

Introduction

Modern imaging techniques have revolutionalised the management of anal canal disorders. Using both ultrasound and MRI, it is possible to acquire exquisitely detailed high-resolution images of the anal sphincter complex and the surrounding tissues. Indeed, imaging has itself contributed significantly to our understanding of normal anal canal anatomy. The role of imaging in the management of anal canal pathology is described elsewhere in this volume. This chapter introduces the techniques of both ultrasound and MRI as applied to the anal canal and highlights both normal appearances and anatomical variations.

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Endoanal Ultrasound

Basic Technique

Before the advent of dedicated endoanal ultrasound, endorectal ultrasound using a 7.5 MHz transducer was being used for rectal cancer staging and prostatic imaging. The transducer was covered with a rubber balloon and this balloon was inflated with degassed water. In 1989, Professor Clive Bartram working at St. Mark's Hospital, London, realised that the technique could be modified to image the anal sphincter complex, simply by implementing a rigid plastic cone so the rotating transducer could be safely withdrawn into the anus.

Since this time, ultrasound transducer technology has progressed, and modern machines are equipped with high-frequency probes providing high-resolution images of the sphincter complex. Isotropic 3-dimensional image acquisition is also increasingly available. The basic technique is however little changed since the original description [1]. The probe is introduced into the anal canal with the patient lying in either the left lateral or prone position. There is evidence that distortion of the anal canal structure is reduced in the prone position, particularly in women [2].

After introduction the probe is slowly withdrawn along the length of the anal canal. Conventionally images are obtained at proximal (puborectalis) level, mid canal and distal level (just below the termination of the internal sphincter). However, as noted above, with many machines it

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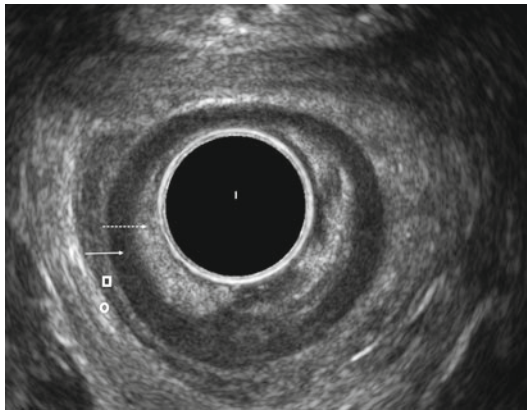

Fig. 3.1 Endoanal ultrasound demonstrating normal sphincter anatomy. Subepithelium (*dotted arrow*) and internal anal sphincter (*thin arrow*). The longitudinal muscle (*square*) and external anal sphincter (*circle*) with comparable reflectivity are distinguished by their interface reflections

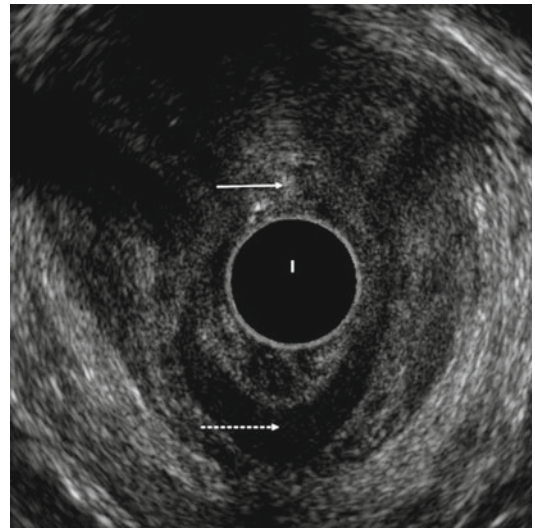

Fig. 3.2 Endoanal ultrasound with an anterior tear of the internal and external anal sphincters (*thin arrow*). Note the retraction of the internal anal sphincter posteriorly (*dotted arrow*)

is now possible to acquire a 3D volume encompassing the entirety of the anal canal. Endoanal ultrasound is usually well tolerated by the patient and can be performed in less than 5 min by an experienced operator. The patient should be reassured that discomfort, if any, will be minimal and short lived.

Patients are requested to lie still and ideally take shallow breaths during the examination if they find it uncomfortable [2].

Normal Anatomy

Accurate interpretation of endoanal US requires a detailed knowledge of anal canal anatomy. The two main sphincters within the canal form two cylindrical layers. The innermost internal anal sphincter (IAS) is composed of smooth muscle, and the outermost external anal sphincter (EAS) is composed of striated muscle. Both are usually well seen on ultrasound. For the purposes of imaging, the anal canal is commonly divided into four main layers: subepithelium, IAS, longitudinal layer/intersphincteric plane and EAS (Fig. 3.1). The validity of ultrasound in defining these discrete layers has been eloquently demonstrated by Sultan et al. using histological dissection of cadaveric specimens [3].

The subepithelium forms the innermost layer. It is mildly hyperreflective (i.e. "bright") and contains the muscularis submucosae ani and vascular channels. The IAS forms the innermost muscular layer and extends from the anorectal junction to approximately 1–1.5 cm below the dentate line. Its characteristic features are of a low-reflectivity (i.e. dark), very well-demarcated ring. As discussed below, the thickness of internal sphincter when measured by ultrasound varies according to age, but it is usually symmetric in the mid canal, although may appear thinner anteriorly, particularly in the upper canal in females. Significant asymmetry in the internal sphincter suggests either focal atrophy or injury (Fig. 3.2). One caveat to this rule however is in the distal canal—an asymmetric termination of the internal sphincter is relatively common in normal individuals which may give the impression of a distal defect to the inexperienced. Conventionally the internal sphincter is measured at its thickest portion at either 3 or 9 o'clock.

The longitudinal muscle interdigitates between the EAS and IAS and terminates in the subcutaneous EAS and subcutaneous peri-anal soft tissues. Sub-adventitial fat on either side of this layer creates interface reflections ("bright lines");

the outer reflection between the longitudinal layer and the external sphincter borders the intersphincteric space. It is postulated that the longitudinal layer functions to brace the anus preventing eversion during defaecation [4]. The ability to clearly depict the longitudinal muscle with ultrasound differs between individual patients but when seen is usually hyporeflective (darker) in comparison to the surrounding echogenic (bright) fat.

The EAS is anatomically composed of three cylindrical bundles. The EAS forms the outermost muscle of the anal canal and extends distally approximately 1 cm beyond the IAS. The deep part is fused to the puborectalis muscle. The EAS is anteriorly related to several identifiable structures including the superficial transverse muscle of the perineum and the perineal body. The transverse perineal muscles usually merge with the anterior EAS in women, but a clear plane is commonly seen between the two in men. Posteriorly the EAS is continuous with the anococcygeal ligament, the low reflectivity of which can sometimes be mistakenly confused for a posterior sphincter defect. Importantly, as discussed below, the EAS is much shorter in women than men [5], and again the inexperienced may confuse this anatomical difference with a sphincter defect. The outer border of the external sphincter is demarcated by the interface reflection with fat in the ischioanal fossa. In cases of fatty atrophy of the external sphincter, this outer border may be difficult to confidently discern on ultrasound, and as discussed below, MRI is superior in assessing EAS atrophy and muscle quality. Indeed, there is good data showing greater interobserver variation in EAS thickness measurement compared to the better-visualised IAS: Gold et al. [6] reported that the 95 % limits of agreement for EAS measurements span 5 mm, compared to just 1.5 mm for the IAS.

As the examination commences in the proximal anal canal, the puborectalis and transverse perineal muscles should be identified. The puborectalis can be distinguished from the EAS as it slings around the anorectal junction and has anterior ends that splay outwards as they point to their insertion at the pubic arch. Once the probe is withdrawn a couple of centimetres, the puborectalis will blend imperceptibly with the EAS and where the sphincter forms a complete ring anteriorly demarcates the mid anal canal. The IAS is also thickest and imaged best at this location. Further withdrawal of the probe images the subcutaneous EAS, and at this level the IAS has usually terminated.

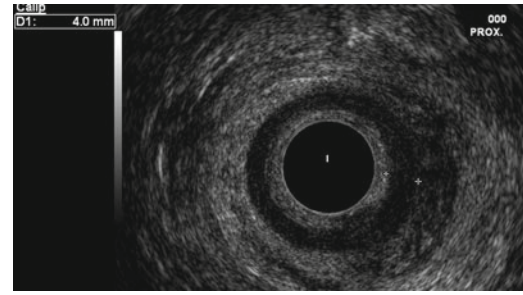

Fig. 3.3 Endoanal ultrasound demonstrating thickened internal anal sphincter (4 mm) in a 50 year old woman (see *calipers*)

Anatomical Variations

There is clear anatomical variation between men and women in anal canal anatomy [5]. The external sphincter is generally shorter in women, particularly anteriorly. Using 3D ultrasound, Williams et al. reported that on average the external sphincter was approximately 17 mm long in women compared to 30 mm in men. The external sphincter is also usually less reflective in men compared to women and therefore easier to visualise. Conversely, little gender differences have been found in the length of either the puborectalis or the IAS.

Sphincter thickness as depicted by endoanal ultrasound is not constant throughout life; notably IAS thickness increases with age. Measurements of 1–2 mm for young adults, 2–3 mm for middle-aged adults and 3–4 mm for the elderly are typical (Fig. 3.3). As the sphincter becomes thicker with age, a small increase in reflectivity may also be noted, attributable by some to a more prominent connective tissue component [7]. Conversely, the external sphincter tends to thin in older nulliparous women, whereas the subepithelium and longitudinal layer tend to remain unchanged [7].

MRI

Basic Technique

In general MRI examination of the anal canal is more complex than endoanal ultrasound. Image quality, for example, can be improved by patient preparation such as a period of fasting to reduce peristalsis artefact, and an empty bladder is preferred.

There are two main approaches to performing anal canal MRI; either the MRI coil is placed over the pelvis of the patient (i.e. externally) or a specially designed endocavity coil is inserted into the anal canal, similar to an endoanal ultrasound probe, although usually of marginally greater diameter. The endoanal coil is introduced with the patient in the left lateral decubitus position, and a supine position is then adopted with the coil secured with cushioning sandbags prior to imaging [8].

Although use of an endocoil is clearly a much more invasive approach impacting on patient acceptability, acquired images have a higher spatial resolution than those produced using an external pelvic coil alone, and highly detailed images can be obtained. However, the field of view (volume of tissue imaged) using an endocoil is restricted—only the anal sphincter complex and a few centimetres of surrounding tissue are well visualised. Conversely the field of view is much greater using an external coil where not only the anal canal is interrogated (albeit with slightly lower but usually adequate spatial resolution) but also the surrounding tissues of the pelvis. Such greater anatomical coverage assumes importance when imaging pathological conditions which often stray beyond the confines of the anal sphincter complex such as peri-anal sepsis and anal cancer. Although many of the early descriptions of anal canal MRI anatomy were based on studies using an endocoil, in clinical practice imaging using the external pelvic coil is overwhelmingly now the preferred option, and the use of endocoils is restricted to a handful of specialist centres.

While there is some debate as to the optimal MR sequence combinations used to image the anal canal, in reality several sequences are useful and choice boils down to the target pathology and individual radiologist preference. T1-weighted IV contrast-enhanced MR imaging, short-inversion-time inversion recovery (STIR) imaging and T2-weighted spin-echo MR imaging are all advocated [9–11] (Table 3.1). A basic T2-weighted fast spin-echo is generally recommended when precise depiction of anal canal anatomy is required since it optimises contrast between the low-signal anal sphincters and the surrounding high-signal fat (see Table 3.1) [12]. Conversely, sequences which null the signal from fat (either fat-saturated T2 or STIR) are particularly useful in depicting peri-anal sepsis when it is important to highlight fluid in tracts, which appear as "bright" high signal. Perhaps the key to successful MRI of the anal canal is optimal slice orientation. It is vital to acquire images carefully aligned to the plane of the anal canal; otherwise, the anatomy is distorted and interpretation much more difficult. Straight axial images through the pelvis are particularly problematic since the anal canal axis is approximately 45° to this. It is paramount therefore to have well-trained radiographic staff supervise the acquisition of anal MRI.

Whether an external pelvic coil or endocoil is used, images are usually acquired in the angled axial and coronal plane, supplemented by sagittal images as required. It is important that the field of view includes the full length of the sphincters and the levators above.

Table 3.1 T2-weighted fast spin-echo sequence recommendations [12]

Parameter	
TR	2,500–3,500 ms
TE	70–90 ms
Echo train length	10
Field of view	10×10 cm (axial) and 16×16 cm (coronal)
Imaging matrix	256×512
Slice thickness	3 mm
Interslice gap	0.3 mm
Excitations	2

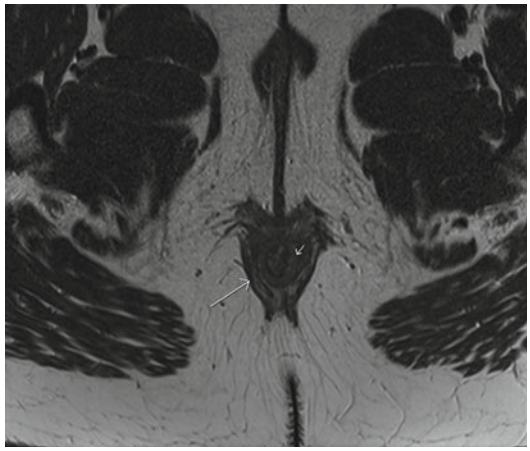

Fig. 3.4 Axial T2-weighted fast spin-echo MRI image of the anus: internal anal sphincter (*small arrow*), external anal sphincter (*large arrow*) and anococcygeal ligament (*dotted arrow*)

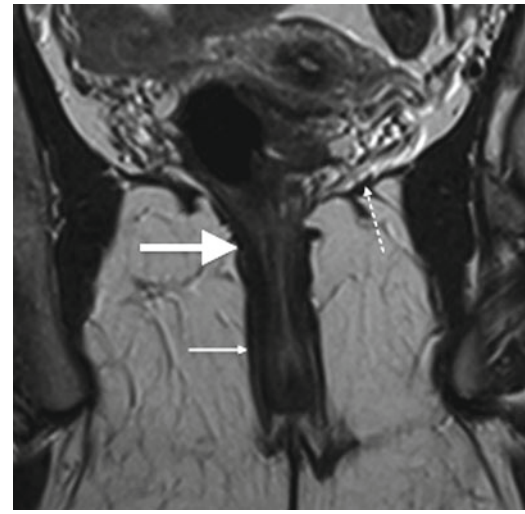

Fig. 3.5 Coronal T2-weighted fast spin-echo MRI image of the anus: external anal sphincter (*thin arrow*), puborectalis (*thick arrow*) and levator ani (*dotted arrow*)

Imaging Anatomy

While much of the anatomy of the anus has already been covered in the discussion regarding endoanal ultrasound, features specific to MRI merit further consideration.

The innermost IAS appears as a relatively hyperintense (i.e. slightly bright) circular structure on T2-weighted images, with a homogeneous uniform architecture (Fig. 3.4). This contrasts with the ultrasound appearances (where the IAS is essentially black). Typical thickness when measured using MRI is approximately 2.9 mm [13], although the previously discussed age variations with US of course still exist with MRI. Atrophy of the internal anal sphincter is best depicted in the axial plane. Abnormal thickening of the anal sphincter, greater than 4 mm, can be found in patients with solitary rectal ulcer syndrome [14]. In contrast, an abnormally thin sphincter, less than 2 mm, can be found in idiopathic degeneration [15].

Contrasting with the IAS, the outermost EAS is a relatively well-defined hypointense (dark) signal structure. A thickness of 4 mm is reported and it extends approximately 1 cm lateral to the IAS. Best imaged in the coronal plane, it can have a characteristic 'J'-shape (Fig. 3.5). Atrophy of the EAS can be accurately depicted using MRI

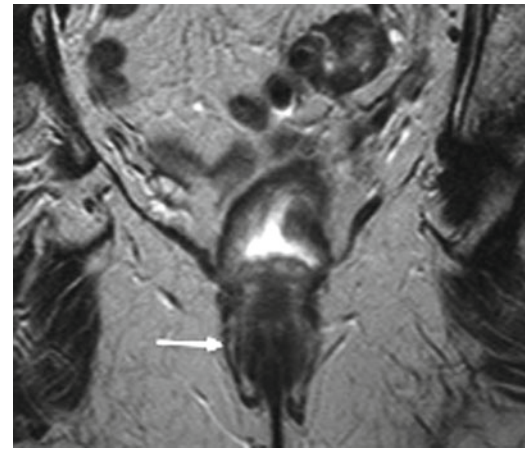

Fig. 3.6 Coronal T2-weighted fast spin-echo MRI image of the anus: note atrophy of the external anal sphincter (*thin arrow*)

(Fig. 3.6). Moderate atrophy has been defined as less than 50 % thinning of less than 50 % normal and/or replacement of the EAS with fat. Severe atrophy is described thinning of 50 % or greater of the usual muscle thickness [16–18].

The longitudinal muscle layer is a relatively hypointense layer on T2-weighted images, lying within the hyperintense intersphincteric space. It is the fibromuscular continuation of the longitudinal muscular layer of the rectal wall.

The puborectal muscle is part of the levator ani complex and forms a sling-like structure, blending into the deep EAS. The MR characteristics are of a relatively hypointense signal intensity structure and it is most simply depicted in the coronal plane [19]. In this plane it is most readily identified as a "budge" at the superior aspect of the deep external sphincter (Fig. 3.5). The high signal returned by the fat in the ischioanal fossa on T2-weighted images facilitates easy identification of the external border of the anal sphincter complex on MRI (superior to ultrasound). As noted above, fat-saturated and STIR sequences are often employed to deliberately null the high signal from the fat to better identify fluid and sepsis. If IV contrast is used, T1-weighted sequences are employed, again usually with fat saturation applied to null signal from fat and allow better appreciation of contrast enhancement. The anatomical detail afforded by MRI allows the observer to appreciate the sheet-like configuration of the external sphincter which may appear "noncontinuous"—a pitfall causing over-diagnosis of sphincter injury by inexperienced observers.

Other anatomical variations are related to patient gender. The transverse perineal muscle also fuses with the EAS in women, while in men it inserts into the central point of the perineum, just as easily appreciated on MRI than on ultrasound. One advantage of external coil MRI is the ability to clearly delineate muscles of the pelvic floor beyond the anal sphincter complex. This may be particularly relevant when imaging the sequelae of obstetric trauma. The puborectalis is seen as a separate structure on MR lateral to the puboviscer-alis complex (puboperineal, pubovaginal and puboanal muscles) and is best depicted in the axial plane where it appears as a relatively symmetric concentric sling, which is hypointense on T2-weighted images. Thinning or defects of the puborectalis can be detected. Asymmetry of the muscle, with the right being thinner than the left, has been described as a normal variant on MRI [20]. The iliococcygeus muscle of the levator ani is well visualised in the coronal plane and is normally convex upwards. Similarly the urogenital diaphragm is best visualised in the coronal plane.

Comparison of MRI and Endoanal US

Both modalities have their strengths and weakness and, as described in later chapters, are used with different frequencies depending on the underlying clinical indication and availability. Endoanal US is cheaper and much quicker than MRI although arguably more observer dependent. It gives very highly detailed images of the sphincter complex and is the investigation of choice for suspected sphincter injury. It has a role in the evaluation of peri-anal sepsis, particularly if simple, but can be painful for patients and may miss unsuspected remote extensions because they are beyond the depth of penetration of the ultrasound beam.

MRI using an external pelvic coil provides a larger field of view of the pelvic tissues, including the pre-sacral space is often employed for the investigation of complex fistula-in-ano and anal cancer staging.

Summary

Imaging the normal anal canal requires a sound understanding of anatomy and has been vastly improved by technological advances over the last two decades. While there are clear indications for the use of ultrasound or MRI as modalities, knowledge of age-related differences and normal variations are crucial if the observer is not to over-diagnose pathology.

References

1. Law PJ, Bartram CI. Anal endosonography: technique and normal anatomy. Gastrointest Radiol. 1989; 14:349–53.
2. Frudinger A, Bartram CI, Halligan S, Kamm M. Examination techniques for endosonography of the anal canal. Abdom Imaging. 1998;23:301–3.
3. Sultan AH, Nicholls RJ, Kamm MA, Hudson CN, Beynon J, Bartram CI. Anal endosonography and correlation with in vitro and in vivo anatomy. Br J Surg. 1993;80(4):508–11.
4. Lunniss PJ, Phillips RK. Anatomy and function of the anal longitudinal muscle. Br J Surg. 1992;79(9): 882–4.

5. Williams AB, Cheetham MJ, Bartram CI, et al. Gender differences in the longitudinal pressure profile of the anal canal related to anatomical structure as demonstrated on three-dimensional anal endosonography. Br J Surg. 2000;87:1674–9.
6. Gold DM, Halligan S, Kmiot WA, Bartram CI. Intraobserver and interobserver agreement in anal endosonography. Br J Surg. 1999;86:371–5.
7. Frudinger A, Halligan S, Bartram CI, Price AB, Kamm MA, Winter R. Female anal sphincter: age-related differences in asymptomatic volunteers with high-frequency endoanal US. Radiology. 2002;224:417–23.
8. Stoker J, Rociu E, Zwamborn AW, Schouten WR, Laméris JS. Endoluminal MR imaging of the rectum and anus: technique, applications, and pitfalls. Radiographics. 1999;19:383–98.
9. DeSouza NM, Kmiot WA, Puni R, et al. High resolution magnetic resonance imaging of the anal sphincter using an internal coil. Gut. 1995;37:284–7.
10. DeSouza NM, Hall AS, Puni R, Gilderdale DJ, Young IR, Kmiot WA. High resolution magnetic resonance imaging of the anal sphincter using a dedicated endoanal coil: comparison of magnetic resonance imaging with surgical findings. Dis Colon Rectum. 1996;39:926–34.
11. DeSouza NM, Gilderdale DJ, MacIver DK, Ward HC. High resolution MR imaging of the anal sphincter in children: a pilot study using endoanal receiver coils. AJR Am J Roentgenol. 1997;169:201–6.
12. Stoker J, Taylor SA, DeLancey JOL. Imaging pelvic floor disorders. 2nd ed. Berlin: Springer-Verlag; 2008.
13. Rociu E, Stoker J, Eijkemans MJ, et al. Normal anal sphincter anatomy and age- and sex-related variations at high-spatial-resolution endoanal MR imaging. Radiology. 2000;217:395–401.
14. Halligan S, Sultan A, Rottenberg G, et al. Endosonography of the anal sphincters in solitary rectal ulcer syndrome. Int J Colorectal Dis. 1995;10:79–82.
15. Vaizey CJ, Kamm MA, Bartram CI. Primary degeneration of the internal anal sphincter as a cause of passive faecal incontinence. Lancet. 1997;349:612–5.
16. Briel JW, Zimmerman DD, Stoker J, et al. Relationship between sphincter morphology on endoanal MRI and histopathological aspects of the external anal sphincter. Int J Colorectal Dis. 2000;15:87–90.
17. Terra MP, Deutekom M, Beets-Tan RG, et al. External anal sphincter atrophy at endoanal MRI in patients with fecal incontinence. Dis Colon Rectum. 2006; 49:1149–59.
18. Terra MP, Beets-Tan RG, Van der Hulst VPM. MR imaging in evaluating atrophy of the external anal sphincter in patients with fecal incontinence. AJR Am J Roentgenol. 2006;187:991–9.
19. Terra MP, Stoker J. Faecal incontinence: endoanal ultrasonography and MR imaging. In: Santoro GA, Di Falco G, editors. Benign anorectal diseases: diagnosis with endoanal and endorectal ultrasonography and new treatment options. Milan: Springer; 2006.
20. Fielding JR, Dumanli H, Schreyer AG, et al. MR based three-dimensional modeling of the normal pelvic floor in women, quantification of muscle mass. AJR Am J Roentgenol. 2000;174:657–60.

Surgical Treatment and Pathology: Normal Histology

4

Ruma Saraswati and Marco Novelli

Anal Canal: Anatomical, Surgical and Pathological Definitions

There are a number of commonly used definitions of the extent of the anal canal. Perhaps the most widely accepted definition is that the anal canal starts at the pelvic floor and finishes at the anal opening [20]. Surgeons usually define the upper limit of the anal canal at the level of the levator ani muscle (at the anorectal angle) and the lower limit at the anal orifice [16]. The surgical anal canal varies from 3.0 to 5.3 cm in length and is on average slightly longer in the female [17]. Pathologists typically define the anal canal as the area lying between the upper and lower borders of the internal anal sphincter [10] whilst anatomists define the anal canal as lying between the dentate line and the anal verge [17]. The anal canal can also be defined histologically based on the types of specialised mucosa present in the anal region (anal transitional zone and anal squamous zone). The surgical and pathological definitions of the anal canal extend more proximally than the anatomical/histological definitions. This results in the surgical and pathological anal canals including a ring of colorectal mucosa at their upper borders [23] which is not present in the anatomical/histological anal canals. This variation in classification leads to some overlap and confusion in terminology particularly in respect to the nomenclature of pathology in the upper anus (e.g. rectal vs anal tonsil, lower rectum vs anal colorectal zone/anal cuff). For classification and staging of anal pathology, both the WHO [14] and TNM [25] systems use the surgical definition of the anal canal.

The anal canal lies in direct continuation with the rectum above and perianal skin below. Perhaps its most important landmark is the "dentate line" a line which runs circumferentially at approximately the midpoint of the anal canal along the base of the anal columns. These columns (Columns of Morgagni) are 6–10 vertical mucosal folds which extend from the middle into the upper aspect of the anal canal. The anal columns are connected at their bases by small semilunar valves behind which there are small pockets called anal sinuses (or anal crypts). The anal glands drain into these sinuses. Below the dentate line, the anal canal has a smooth lining and this area is referred to as the pecten.

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Basic Histological Structure of the Anus

The anus has a similar structure to the rest of the gastrointestinal tract being composed of mucosa, submucosa and a muscular wall. However, unlike other parts of the large bowel, the muscular wall

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has a component of striated (voluntary) muscle (external anal sphincter) in addition to the usual muscularis propria (internal anal sphincter). There is marked variation in the mucosal histology within the anus (see section "Histological zones of the anus" and Fig. 4.1). The submucosa of the upper anus is similar to the submucosa of the rectum being composed of loose connective tissue, but in addition it contains a rich venous plexus (see vascular supply). Below the dentate line in the pecten, there is more dense fibroblastic tissue which tethers the squamous epithelium to the superficial aspect of the internal sphincter. This is said to form a submucosal barrier which may be important in limiting the spread of carcinoma.

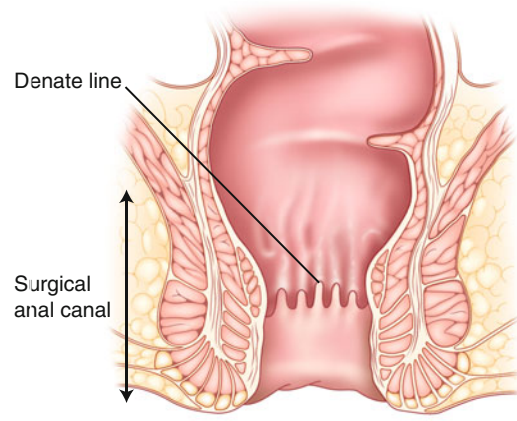

Fig. 4.1 Histological zones of the anal canal

Histological Zones of the Anus

The anal canal is divided into three separate histological zones according to the type of lining mucosa: the colorectal zone, the anal transitional zone and the squamous zone [9, 11]. These zones can be roughly mapped onto the anatomical landmarks of the anus, but there is some variation both between and within individuals [21].

The Colorectal Zone

The anal colorectal zone is lined by large intestinal-type mucosa and blends imperceptively with the lower rectal mucosa. The mucosa of the colorectal zone typically shows more crypt architectural distortion and shorter crypts than more proximal large intestinal mucosa with frequent branched/bifid crypts, and there are often mild mucosal prolapse-type features in this area (these include mild angulation of crypt outlines and mild fibromuscularisation of the lamina propria with vertically orientated bands of smooth muscle extending up between crypts). There are also changes in the type of mucin with goblet cells of the anorectum predominantly producing sialomucins rather than the sulphomucins found in the colorectum [6]. The colorectal zone starts at around the level of puborectalis and is typically 1–2 cm in length. Its mucosa merges with the mucosa of the anal transitional zone above, but close to, the dentate line.

The Transitional Zone

The anal transitional zone is lined by transitional-type mucosa and merges with the squamous zone below. There is an abrupt change from transitional- to colonic-type mucosa at the junction with the colorectal zone above. In some areas the transitional zone may be absent with large intestinal-type mucosa of the colorectal zone directly abutting squamous mucosa of the squamous zone. Other names for the transitional zone include the "intermediate zone" and the "cloacogenic zone". The anal transitional zone typically starts just above the dentate line, approximately 10 mm above the lower border of the internal sphincter, and is on average 5 mm in length, ranging between 3 and 11 mm [21]. The anal transition zone contains anal ducts which connect with the anal glands (see The Anal Blood Supply).

The anal transitional epithelium is normally 4–9 cell layers thick. The basal cells are small and perpendicularly arranged to the basement membrane. Cells at the surface of the anal transi-

tional mucosa can be columnar, cuboidal, flattened or polygonal. The columnar cells produce small volumes of mucin. Foci of squamous differentiation and occasional colonic-type glands may also be present. Endocrine cells are scattered in the transitional epithelium, and occasional melanocytes may be present [11] but Langerhan's cells are absent [12]. Similar to transitional epithelium in other regions, such as the bladder, anal transitional epithelium stains positively with CK 19 [24]. The transitional zone merges with the anal squamous zone below typically just above the level of the dentate line.

The Squamous Zone

The anal squamous zone (sometimes referred to as the "smooth" zone) anatomically corresponds to the pecten of the anal canal [10]. It is lined by non-keratinising squamous epithelium which merges at its lower border with perianal skin. The squamous zone lacks well-formed dermal papillae and skin appendages/adnexal structures. The squamous zone also lacks mucin-producing cells but melanocytes are present. As the epithelium approaches and merges with perianal skin, there is a gradual change in its cellular constituency with an increase in the number of melanocytes [4] and Langerhan's cells [12].

Mucosa-Associated Lymphoid Tissue of the Anus

There is mucosa-associated lymphoid tissue distributed throughout the large intestine but the amount varies markedly depending on the location. Lymphoid follicles are seen in the mucosa of the anal canal from the dentate line upwards [15].

In the anorectal region, the mucosa-associated lymphoid tissue can form a polypoidal structure which is known as a rectal tonsil [8]. This is more commonly seen in young children and adolescents. The aetiology of this is uncertain in most cases but the literature suggests that some of these cases may be linked to anorectal infections such as chlamydia [5]. Knowledge of this "normal" structure is important to avoid its misdiagnosis as lymphoproliferative disease.

The Anal Glands and Ducts

The anal glands drain via ducts which open into the anal transitional zone. Approximately 80 % of anal glands lie in the submucosa; however, the remainder of the anal glands are situated more deeply in the anal wall within the internal sphincter, the intersphincteric space and even the external sphincter [19]. Infection within anal glands is considered to be the main aetiological factor in the development of perianal fistulae [18].

The epithelial lining of anal glands is very similar to that of the anal transitional zone but a characteristic feature of the epithelium is the presence of intra-epithelial microcysts [11].

The Anal Musculature

The anal muscular wall is composed of the internal and external anal sphincters. The internal anal sphincter is a continuation of the muscularis propria of the rectum to which it has a similar structure although it is considerably thicker. The external anal sphincter is formed of striated muscle, is said to be composed of superficial and deep components and surrounds the internal sphincter.

The Anal Blood Supply

The anus has a rich arterial blood supply which is predominantly derived from the superior rectal artery (a direct continuation of the inferior mesenteric artery) but also receives contributions from the middle and inferior rectal arteries (branches of the internal iliac/pudendal arteries). In the upper anal canal, above the dentate line,

the anal submucosa contains a rich vascular plexus known as the anorectal vascular plexus which includes numerous arteriovenous connections. This is not evenly distributed throughout the upper anal canal but concentrated to form three anal cushions (left lateral, right anterior and right posterior) which have erectile tissue-type properties and are thought to be involved in the maintenance of continence. The anal cushions are also thought to be important in the pathogenesis of haemorrhoids [1, 22].

The Anal Nerve Supply

The inferior rectal nerves supply sensory innervation to the lower part of the anal canal. Free and organised nerve endings extend throughout the squamous zone and up into the transitional zone finishing just above the dentate line and providing sensation to touch, pain, hot and cold. However, above this the colorectal zone is devoid of such nerve endings and is dull to sensation [7, 11].

In the upper anal canal, as in the colorectum, the autonomic innervation is provided through submucosal and myenteric plexi which contain numerous ganglion cells. However, for the first centimetre above the dentate line (corresponding to the anal transitional and lower colorectal zones), ganglion cells are sparse or absent [2]. The density of interstitial cells of Cajal (the pacemaker cells of the colorectum) is also markedly reduced in the internal anal sphincter as compared to the rectum [13].

The Anal Lymphatic Supply

Injection studies have shown that an indirect or intramural system connects the lymphatics of the upper part of the anal canal to the lymphatic plexuses of the rectum and then onto the upper rectal nodes [3]. The lower part of anal canal below the dentate line and perianal skin is drained by the superficial inguinal nodes.

References

1. Aigner F, Gruber H, Conrad F, Eder J, Wedel T, Zelger B, Engelhardt V, Lametschwandtner A, Wienert V, Böhler U, Margreiter R, Fritsch H. Revised morphology and hemodynamics of the anorectal vascular plexus: impact on the course of hemorrhoidal disease. Int J Colorectal Dis. 2009;24(1):105–13.
2. Aldridge RT, Campbell PE. Ganglion cell distribution in the normal rectum and anal canal. A basis for the diagnosis of Hirschsprung's disease by anorectal biopsy. J Pediatr Surg. 1968;3(4):475–90.
3. Blair JB, Holyoke EA, Best RR. A note on the lymphatics of the middle and lower rectum and anus. Anat Rec. 1950;108(4):635–44.
4. Clemmensen OJ, Fenger C. Melanocytes in the anal canal epithelium. Histopathology. 1991;18(3):237–41.
5. Cramer SF, Romansky S, Hulbert B, Rauh S, Papp JR, Casiano-Colon AE. The rectal tonsil: a reaction to chlamydial infection? Am J Surg Pathol. 2009;33(3):483–5.
6. Fenger C, Filipe MI. Mucin histochemistry of the anal canal epithelium. Studies of normal anal mucosa and mucosa adjacent to carcinoma. Histochem J. 1981;13(6):921–30.
7. Duthie HL, Gairns FW. Sensory nerve-endings and sensation in the anal region of man. Br J Surg. 1960;47:585–95.
8. Farris AB, Lauwers GY, Ferry JA, Zukerberg LR. The rectal tonsil: a reactive lymphoid proliferation that may mimic lymphoma. Am J Surg Pathol. 2008;32(7):1075–9.
9. Fenger C, Lyon H. Endocrine cells and melanin-containing cells in the anal canal epithelium. Histochem J. 1982;14(4):631–9.
10. Fenger C. The anal transitional zone. Acta Pathol Microbiol Immunol Scand Suppl. 1987;289:1–42.
11. Fenger C. Histology of the anal canal. Am J Surg Pathol. 1988;12(1):41–55.
12. Fenger C, Schrøder HD. Neuronal hyperplasia in the anal canal. Histopathology. 1990;16(5):481–5.
13. Hagger R, Gharaie S, Finlayson C, Kumar D. Distribution of the interstitial cells of Cajal in the human anorectum. J Auton Nerv Syst. 1998;73(2–3):75–9.
14. Hamilton SR, Aaltonen LA. Pathology and genetics. Tumours of the digestive system, WHO classification of tumours, vol. 2. Lyon: IARC WHO Classification of Tumours, No 2; 2000.
15. Langman JM, Rowland R. Density of lymphoid follicles in the rectum and at the anorectal junction. J Clin Gastroenterol. 1992;14(1):81–4.
16. Morgan CN, Thompson HR. Surgical anatomy of the anal canal with special reference to the surgical importance of the internal sphincter and conjoint longitudinal muscle. Ann R Coll Surg Engl. 1956;19(2):88–114.
17. Nivatvongs S, Stern HS, Fryd DS. The length of the anal canal. Dis Colon Rectum. 1981;24(8):600–1.

18. Parks AG. Pathogenesis and Treatment of Fistula-in-Ano. Br Med J. 1961;1(5224):460–2, 463–9.
19. Seow-Choen F, Ho JM. Histoanatomy of anal glands. Dis Colon Rectum. 1994;37(12):1215–8.
20. Symington J. The rectum and anus. J Anat Physiol. 1888;23(Pt 1):106–15.
21. Thompson-Fawcett MW, Warren BF, Mortensen NJ. A new look at the anal transitional zone with reference to restorative proctocolectomy and the columnar cuff. Br J Surg. 1998;85(11):1517–21.
22. Thomson WH. The nature of haemorrhoids. Br J Surg. 1975;62(7):542–52.
23. Walls EW. Observations on the microscopic anatomy of the human anal canal. Br J Surg. 1958;45(193):504–12.
24. Williams GR, Talbot IC, Northover JM, Leigh IM. Keratin expression in the normal anal canal. Histopathology. 1995;26(1):39–44.
25. Wittekind CH, Greene FL, Hutter RVP, Sobin LH, Klimpfinger M. TNM atlas: illustrated guide to the TNM classification of malignant tumours. Hoboken: Wiley-Liss; 2005.

Congenital Abnormalities of the Anus

5

Nigel J. Hall and Edward M. Kiely

Introduction

Congenital anorectal abnormalities occur with a frequency in the region of 1:5,000 live births and form a spectrum of disorders in which the anatomy of the rectum, anal canal and anus is malformed. The resulting clinical phenotype ranges from a stenosed anus in an anatomically normal or near-normal position, through an abnormal fistulous connection between the distal end of the gastrointestinal tract and part of the genitourinary tract, to the presence of a long common channel (a persistent cloaca) into which colon, bladder and vagina all open into a single cavity with a single external opening.

To correct these abnormalities requires a detailed understanding of the anatomy of the pelvic floor and perineum. The aim of surgery is to provide a normal functional outcome in terms of urinary voiding and renal function, continence and defecation and sexual function. Due to the complexity of some of these abnormalities and the high incidence of associated congenital anomalies, achieving good functional outcome is challenging.

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The cause of congenital anorectal abnormalities is unknown. In affected infants and children, there is usually no family history of such anomalies and low risk to succeeding generations. Uncommonly, there are families where there is a genetic predisposition, whose mode of inheritance is unclear except for the isolated example of the Currarino triad. In this triad – anal anomaly, sacral anomaly, presacral mass – the chromosomal abnormality has been defined. Anorectal abnormalities may be seen with increased frequency in Down's syndrome, in association with oesophageal atresia and in those with Hirschsprung's disease. The genetic defect in these infants has not been defined. There is an excess of affected males (55–65 %) which is unexplained. The gender distribution of abnormalities is also unequal; over half of affected males have high, supralevator lesions and over half of affected females low, infralevator lesions.

Embryology

The older theories on anorectal development have given way to more recent information gleaned from studies on different mammals. These newer studies have used microdissection and scanning electron microscopy to provide three-dimensional information on both normal and abnormal anorectal development. The animal models used have included rodents and pigs bred for their susceptibility to anorectal malformations. The development of the anorectum is

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very similar in these different species, and the anorectal malformations they exhibit are similar to those found in humans.

In the early weeks of human development, the hindgut and eventually the urinary tract enter a common cavity – the cloaca. This cavity is closed to the outside by the cloacal membrane which extends from the body stalk superiorly to the tail inferiorly. The cloaca is formed by the third week of gestation.

A urorectal septum divides the cloaca into anterior and posterior segments and eventually reaches the cloacal membrane which then disintegrates. Two openings are then established – urogenital anteriorly and anal posteriorly. This process is completed by about the seventh week of gestation. During this time, the mucosal lining of anterior and posterior cloacas is closely applied to the epithelium of the cloacal membrane. In animal models with anorectal malformations, the cloacal membrane is deficient posteriorly and the posterior cloaca is much shorter than normal. Finally, mesoderm is interposed between the cloacal epithelium externally and cloacal mucosa internally.

The future site of the anus is clearly seen from the earliest weeks at the dorsal (inferior) extent of the cloacal membrane. In normal development, there is no mesoderm between the cloacal cavity and the cloacal membrane at this level. When the cloacal membrane breaks down, normal anatomy is established.

In the presence of an anorectal malformation, the anus is misplaced and often stenosed. The communication of hindgut to anterior cloaca persists, and in postnatal life, this opening has the physical features of an anus – mucosal lining and internal sphincter. At the present time, the development of the frequently associated genitourinary abnormalities is unexplained.

Classification

A number of different classifications for anorectal malformations have been proposed over the years. One such system previously used divided anomalies into high, intermediate and low based on the distance of the tip of the rectal stump from the perineal skin. In infants with high anomalies, the bowel terminates above the pelvic floor. In those with low anomalies, the bowel traverses the levator muscle and the anus is then ectopic and/or stenotic. Inevitably, there is a small number who do not fit into either group and these are termed intermediate anomalies. Proponents of such a system suggested that this facilitated comparison of results of treatment as, in broad terms, the functional outcome of anorectal malformation is related to the 'height' of the anomaly. However, such a system does not take into account the fine anatomical detail of the anomaly that is required for precise surgical repair. We therefore prefer a descriptive classification based on the anatomical details (Table 5.1).

Male Defects

Rectoperineal Fistula (Fig. 5.1)

This is the lowest of the male defects. There is an opening of a fistula onto the perineum usually in the midline and almost always anterior to the sphincter mechanism. The opening may be as far anterior as the base of the scrotum and in some cases on the scrotum itself. Often the baby passes meconium through the fistula although this may take several hours following birth. The perineum is otherwise usually normal, and the likelihood of having an associated spinal or genitourinary

Table 5.1 Classification (in order of decreasing frequency)

Male defects
Rectourethral fistula
Rectoperineal fistula
Rectovesical (bladder neck) fistula
Imperforate anus with no fistula
Rectal atresia/stenosis
Female defects
Rectovestibular fistula
Rectoperineal fistula
Cloacal malformation
Imperforate anus with no fistula
Rectal atresia/stenosis

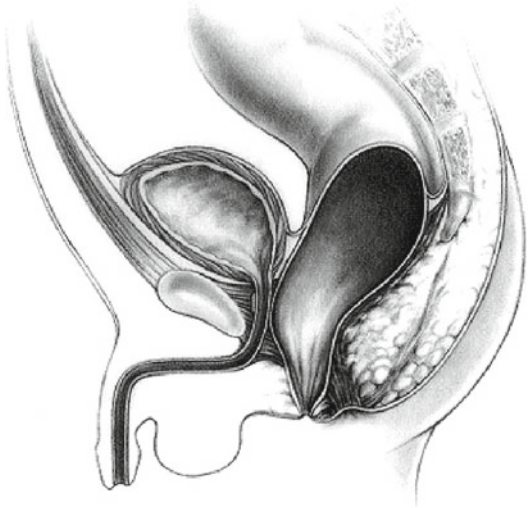

Fig. 5.1 Rectoperineal fistula (Figure 47.4, page 464 from *Pediatric Surgery: Diagnosis & Management*, Eds: Puri & Hollwarth, Springer 2009: ISBN 978-3-540-69559-2)

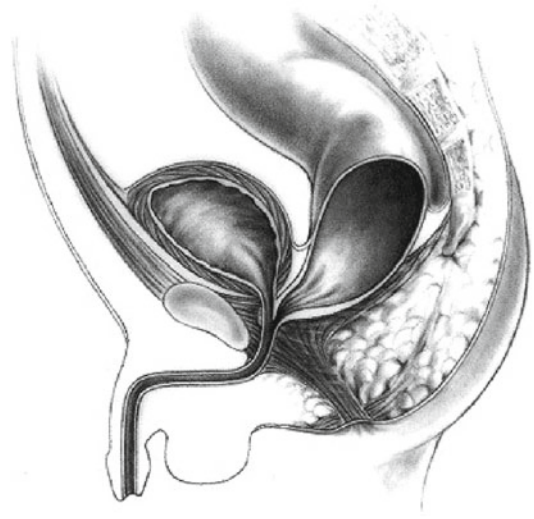

Fig. 5.2 Rectoprostatic fistula (Figure 47.2, page 464 from *Pediatric Surgery: Diagnosis & Management*, Eds: Puri & Hollwarth, Springer 2009: ISBN 978-3-540-69559-2)

abnormality is low. In the absence of an obvious opening or the appearance of meconium on the perineum, other clinical features that suggest the presence of this low type of malformation include a bucket-handle-type deformity or a subepithelial strand of white mucous. Selected cases may be repaired with a minimal anorectoplasty in the newborn period, thereby avoiding the need for colostomy.

Rectourethral Fistula (Fig. 5.2)

The characteristic presentation of this type of defect is the absence of an anus with the appearance of meconium or meconium staining in the urine. There may be an anal dimple, well-developed buttocks and a midline groove with a bulbar urethral fistula but the chance of having such a 'normal' appearing perineum are lower with a fistula that opens into the prostatic urethra or higher. Internally, there is a common wall between the anterior wall of the fistula and the posterior wall of the urethra. Approximately one-third of cases with a bulbar urethral fistula have an associated genitourinary abnormality and this increases to two-thirds of cases with a prostatic fistula. All these infants require a diverting colostomy followed by a formal posterior sagittal anorectoplasty (PSARP) when the infant is a few months of age.

Rectovesical Fistula (Fig. 5.3)

The fistula usually enters the bladder at the bladder neck and there may be meconium in the urine. The perineum is usually poorly developed with flat buttocks and underdeveloped muscles. There is a high incidence of associated urological and spinal anomalies. Internally the fistula enters the bladder most commonly in a T-shaped orientation without a long common wall. All infants with this high type of anomaly require a diverting colostomy followed by reconstruction when a few months older. The fistula is normally divided laparoscopically and the procedure completed from above rather than the usual approach from below.

Imperforate Anus Without Fistula

This is an uncommon anomaly (5 % of all male defects) and is commonly associated with Down's syndrome. There is no meconium on the

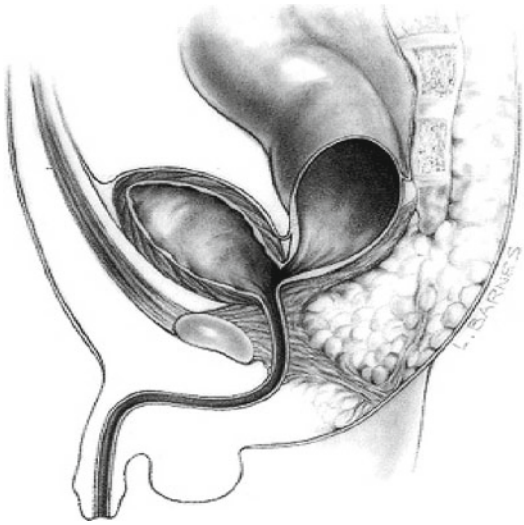

Fig. 5.3 Recto-bladder neck fistula (Figure 47.1 page 464, from *Pediatric Surgery: Diagnosis & Management*, Eds: Puri & Hollwarth, Springer 2009: ISBN 978-3-540-69559-2)

perineum nor in the urine, and the perineal muscle development is often reasonable. There is a low incidence of coexisting anomalies. These infants require colostomy formation in the newborn period followed by PSARP. Despite the absence of a fistula, there may be a long common wall between the rectal stump and the urethra which requires meticulous and complete dissection.

Rectal Atresia/Stenosis

These are the most rare of all anorectal anomalies and the least readily detected. Externally the anus appears entirely normal. Those with an atresia have a normal anal canal, an atresia 1–2 cm above the anal verge and a dilated blind pouch of proximal colon. In the case of rectal stenosis, there is a narrow, often fibrous passage between dilated colon and the anal canal. Both rectal atresia and rectal stenosis may be associated with a presacral mass as part of the Currarino triad. The perineum usually appears normal, but due to the dilated proximal colon, decompressive colostomy is recommended followed by later reconstruction. Because the anal canal is normal, functional outcome is generally good.

Female Defects

Rectovestibular Fistula (Fig. 5.4)

This, the most frequent defect seen in females, comprises a fistula opening into the posterior margin of the vaginal vestibule. The infant is usually able to pass some meconium through the fistula but not an adequate amount to allow decompression. The perineum is usually reasonably well developed, and the incidence of urogenital anomalies is approximately one-third, whilst sacral anomalies are rare. There is usually a long common wall between the fistula and the posterior wall of the vagina which requires meticulous and complete separation during anorectoplasty. The safest approach to an infant with a rectovestibular fistula is to form a colostomy. However, in a selected group of girls with this type of anomaly and an experienced surgeon, it may be possible to dilate the fistula adequately to allow decompression of the bowel prior to performing a primary PSARP without a covering stoma. This may be performed in the newborn period or, more easily, at a few months of age.

Rectoperineal Fistula

The female equivalent of this type of anomaly shares the features of its male counterpart. The fistula opens onto the perineum somewhere between the anal dimple and the vestibule and may often resemble a normal anus in appearance to the extent that some call this anomaly an ‘anterior ectopic anus’. Some cases do not require any surgical intervention so long as the calibre of the anus is adequate and the infant passes stool without undue difficulty. However, when constipation is problematic, a limited anorectoplasty without covering stoma may be performed.

Persistent Cloaca (Fig. 5.5)

Cloacal malformations are the most complex of all the congenital anorectal malformations.

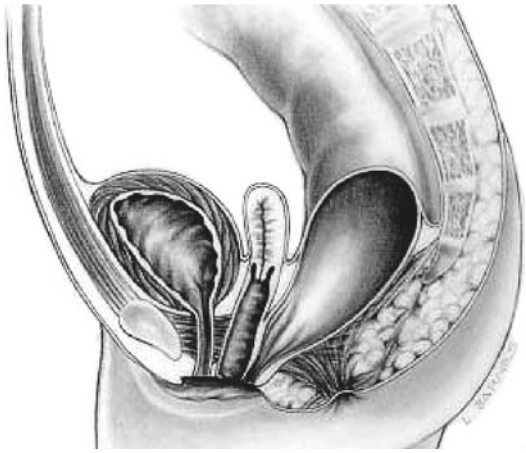

Fig. 5.4 Rectovestibular fistula (Figure 47.8, page 465 from *Pediatric Surgery: Diagnosis & Management*, Eds: Puri & Hollwarth, Springer 2009: ISBN 978-3-540-69559-2)

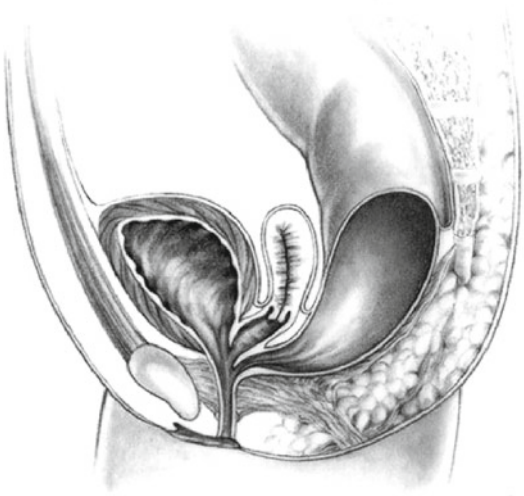

Fig. 5.5 Cloaca with common channel longer than 3 cm (Figure 47.6, page 465 from *Pediatric Surgery: Diagnosis & Management*, Eds: Puri & Hollwarth, Springer 2009: ISBN 978-3-540-69559-2)

The vagina, urethra and rectum all open into a single common channel of variable length. This anomaly represents approximately 10 % of all anorectal malformations in females. There is a spectrum of severity determined primarily by the length of the common channel. This determines complexity of operative difficulty and functional outcome. With a short cloaca (defined as <3 cm in length), prognosis is generally good. However, in the presence of a channel >3 cm in length, there is a high incidence of vaginal and uterine anomalies, sacral anomalies and urinary tract obstruction. A common finding is vaginal obstruction resulting in hydrocolpos which may present as a palpable abdominal mass. The most significant morbidity associated with cloacas is that of obstructive uropathy with up to 30 % progressing to chronic renal failure.

All infants with cloacal malformations require a diverting colostomy and assessment of the urinary tract to ensure that drainage is adequate. On occasion a vesicostomy will be necessary. Hydrocolpos is treated with a vaginostomy. Formal repair occurs during the first year with a posterior sagittal anorectovaginourethroplasty (PSARVUP). For very high defects, an abdominal approach is required in addition to a perineal approach.

Other Female Defects

Imperforate anus without fistula and rectal atresia/stenosis are similar to their male counterparts and treated as such. Some classification systems include the female defect of rectovaginal fistula. In one large series of cases originally diagnosed as having a rectovaginal fistula and requiring reoperation, none were felt to actually have had such an anomaly. Two-thirds had features consistent with a diagnosis of persistent cloaca and the remaining third had a rectovestibular fistula.

Adequate and thorough neonatal assessment of all affected infants (including examination under anaesthetic if necessary) is essential to avoid erroneous surgical treatment with potentially adverse functional outcome. The key clinical features of a rectovaginal fistula are a clearly visible urethral orifice, a normally appearing vaginal orifice and meconium appearing from inside the vagina above the hymen. All such infants require initial colostomy. Detailed investigation and later reconstruction are performed when the baby is older.

Clinical Presentation and Diagnosis

The majority of anorectal anomalies present in the newborn period are detected at or soon after birth or during routine postnatal examination. The absence of an anus or an abnormal appearance of the perineum should draw attention to the underlying malformation. Surprisingly, however, it is not uncommon for the diagnosis to be missed and the infant to present with abdominal distension at a few days of age.

In addition to the abnormal external appearances, clinical features include absence of or minimal passage of meconium together with abdominal distension. In extreme circumstances, infants may present with intestinal perforation or urinary obstruction.

The diagnosis of an anorectal malformation is made on clinical grounds. In the majority of cases, adequate information can be achieved clinically to determine the likely anatomy and whether or not a colostomy is required. It is usual to wait 24 h before making any decision about surgical treatment. This allows time for meconium to appear at the perineum if this is possible. Some will require examination under anaesthesia in order to make a complete assessment.

In infants in whom the anatomy cannot be determined on clinical grounds (usually those in whom no meconium has been seen), some surgeons recommend taking a cross table lateral X-ray with the infant in the prone position (a so-called invertogram) once the child is 24 h of age. The position of the tip of the rectal stump on an invertogram may be seen as an air bubble. The proximity of this bubble to the perineal skin (which is marked using a radio-opaque marker) is used to guide subsequent management. If it is less than 1 cm, it is likely that there is an undiagnosed rectoperineal fistula that the defect is 'low' and a PSARP without colostomy may be possible. A bubble more than 1 cm from the perineal skin indicates a higher anomaly necessitating colostomy. If there is any doubt, the safest option is to perform a colostomy.

Our own practice is to wait the full 24 h, make a detailed examination and then proceed depending on the clinical findings. If there is no meconium on the perineum, we would perform a stoma. If the findings suggest a low anomaly – meconium at skin level – we would aim to perform a local procedure. We have not found the invertogram helpful, as the position of the air bubble is determined by the state of contraction of the pelvic floor. Because of this, the rectum may appear to be much lower than it is and an injudicious attempt may be made at locating the rectum from below. Considerable tissue destruction may then result.

Associated Anomalies

The overall incidence of associated anomalies in infants with anorectal malformations is 50–60 %. The commonest of these involve the genitourinary tract and the spine. An anorectal anomaly may exist as part of the VACTERL association (Vertebral, Anorectal, Cardiac, Tracheoesophageal, Renal and Limb anomalies). In general terms, the higher the anorectal malformation (i.e., the greater the distance between the tip of the rectal stump and the perineal skin), the greater the incidence of associated anomalies.

In order to detect these anomalies, we recommend that all infants born with an anorectal malformation undergoing diagnostic imaging of the renal tract (ultrasound) and spine (X-ray and ultrasound). Further imaging such as micturating cystography or isotope studies can be obtained as necessary depending on the abnormalities found. The most significant associated anomaly in terms of long-term morbidity is an associated cardiac malformation. Tethering of the spinal cord (present in up to 25 % of cases) and anomalies of the genitourinary may affect the long-term bowel and bladder control.

Treatment

The stages of treatment when faced with an infant with an anorectal malformation are shown in Fig. 5.6. As described, the anatomical arrangement of some defects mandates a diverting stoma, whereas some 'low' defects may be

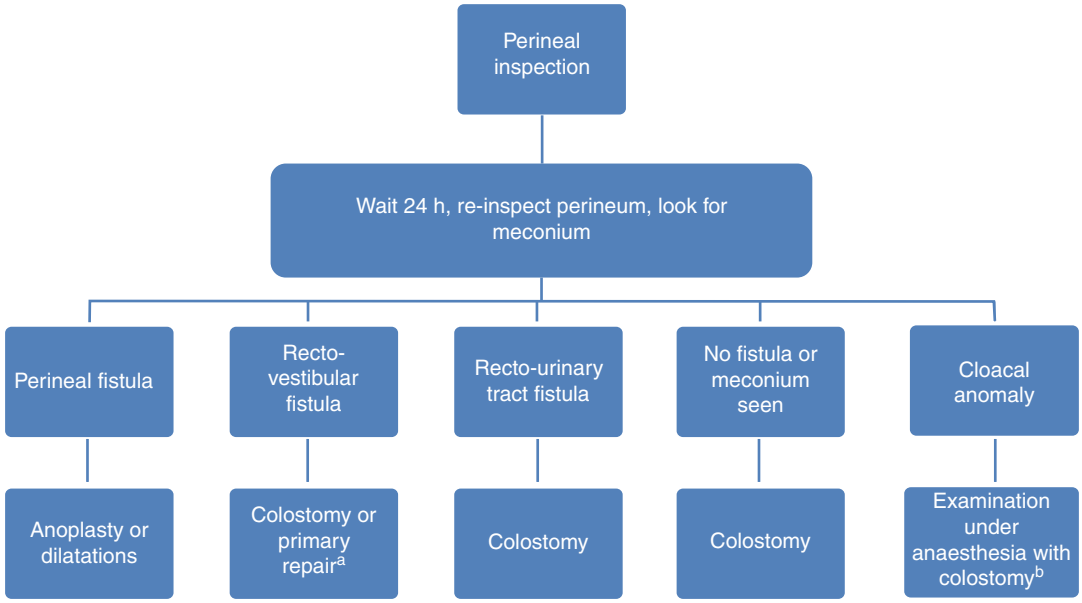

Fig. 5.6 Initial evaluation and treatment of a newborn with an anorectal malformation. [a]Depending on experience of the surgeon and overall condition of the infant. [b]Including urinary tract drainage/diversion and drainage of hydrocolpos as necessary

treated primarily with a perineal procedure. This will depend on the degree of intestinal obstruction and the experience of the surgeon.

When a stoma is required, there is little consensus on the best form of stoma. The options include loop and split colostomies, which are positioned in the transverse, descending or sigmoid colon. The commonest performed is a split sigmoid stoma, but this may not allow sufficient length distally to perform the reconstruction.

The author's preference is to perform a loop proximal transverse colostomy in all patients with an anorectal malformation requiring a stoma. The period of anaesthesia for colostomy formation provides an excellent opportunity for examination under anaesthesia of the perineum to gain further detail of the anatomical arrangement of the defect. This may include cystoscopy and vaginoscopy, particularly in cases of suspected persistent cloaca in which the length of the common channel should also be assessed.

Having achieved safe intestinal decompression, the child can be established on enteral feeds and attention paid to identification of potential associated anomalies. It is our standard practice for all infants to undergo a renal tract ultrasound and an ultrasound and anteroposterior radiograph of the sacral and lumbar spine. Any anomalies detected are further investigated and treated as necessary. Once the infant established on full enteral feeds and the stoma functioning, they are discharged to home. Most infants gain weight adequately with a colostomy. A small number however may have watery stoma output and lose large amounts of sodium in the effluent and consequently have poor weight gain. A urinary sodium concentration of less than 10 mmol/L indicates total body sodium deficiency and that the infant is sodium deplete. Sodium supplementation should be commenced.

Infants with a low defect who have undergone a primary procedure without colostomy can be established on enteral feeds soon after surgery. They are discharged home once the diagnostic imaging has been completed and bowel function is established.

All infants with a colostomy require further imaging of their defect prior to planning formal reconstruction. This achieved by means of a micturating cystourethrogram (MCUG) and a

contrast study of the distal loop of the colostomy (distal loopogram), performed simultaneously. Both distal loop of colon and bladder should be well distended during the studies. Whilst the majority of fistulae are detected, a small proportion cannot be demonstrated and only become apparent during reconstruction. Anorectal reconstruction is performed at around 3–6 months of age.

Colostomy Formation

General anaesthesia is necessary and prophylactic antibiotics are given. The site for the colostomy is chosen to ensure there will be adequate distance around the stoma for attachment of the bag. A V-shaped incision is made and a full thickness skin flap raised, exposing the underlying muscle. The muscle is split transversely, perpendicular to the V incision, and adequately to allow the passage of the loop of colon. The peritoneum is then opened in the same direction as the muscle.

Essential to the success of colostomy formation is identification of the correct part of the intestine and ensuring that it is orientated correctly within the incision. This is particularly true of sigmoid colostomies in infants as the dilated sigmoid colon is very mobile and may be encountered almost anywhere within the abdominal cavity. The presence of an omental attachment differentiates transverse colon from the remainder. The selected loop of bowel is drawn to the surface, ensuring that it is not twisted. The omentum is cleared from the transverse colon. A small opening is created in the colonic mesentery close to the bowel wall, the apex of the V skin flap passed through the mesenteric defect and the skin flap sutured to its original position with two or three loosely tied interrupted sutures.

The colon is then incised longitudinally with cutting diathermy and excess meconium evacuated. Some surgeons recommend flushing out the meconium from the distal limb with normal saline. The colonic mucosa is everted slightly and sutured to the surrounding skin with interrupted absorbable sutures.

Post-operatively, the stomas function rapidly and a colostomy bag can then be applied.

Anorectal Reconstruction

The precise procedure required is determined by the anatomy of the anorectal anomaly. In the lowest of defects comprising either a rectoperineal fistula or an anterior ectopic anus, a relatively minor procedure, usually an anoplasty, may be all that is required. In higher defects we recommend the posterior sagittal anorectovaginourethroplasty (PSARP); for cloacas in which some form of mobilisation of the urogenital tract is necessary, the procedure is the posterior sagittal anorectovaginourethroplasty (PSARVUP). Whilst the majority of anomalies may be repaired through a perineal incision, high defects including those with a rectovesical fistula or a cloaca with a long common channel may require an abdominal approach to achieve adequate mobilisation. All such operations are covered with prophylactic antibiotics.

Anoplasty

This is generally performed when the anus is correctly sited but stenotic. The simplest and most satisfactory is the Y-V anoplasty. A V-shaped incision is made with the apex of the V at the 6 o'clock position. A vertical incision is then made posteriorly up into the anal canal for 1.5–2 cm. The V flap is mobilised and advanced into the anal incision and sutured in position with fine absorbable sutures. No post-operative dilatations are necessary.

Posterior Sagittal Anorectoplasty (PSARP)

This technique can be used to repair the majority of anorectal malformations. In low abnormalities such as a rectoperineal fistula, a limited PSARP may be performed in which the incision is of shorter length and a less invasive dissection is

required. In cases of rectourethral fistula, the incision extends from anterior to the muscle complex to the lower sacrum and the dissection is deeper to allow identification and division of the fistula and adequate mobilisation of the rectum. In high anomalies such as a recto-bladder neck fistula or a high prostatic fistula, a combined approach using laparoscopy is necessary to first divide the fistula and mobilise the rectum prior to formal PSARP utilising a perineal approach.

The principles of the PSARP are to identify and safely divide the fistula, to provide adequate mobilisation to the rectum to place it within the centre of the muscular sphincter complex without tension and finally to perform an anoplasty by suturing the neoanus to the skin.

The PSARP is performed with the infant prone, with hips flexed, slightly abducted and externally rotated. Adequate support is provided to the rest of the infant with soft padding under chest and pelvis to avoid pressure during the procedure. Prior to positioning, the urethra is catheterised. Prior to making the incision, the position of the anal sphincter complex is identified using an electrical stimulator. In all situations the incision is in the midline through the sphincter complex, staying precisely in the midline leaving identical amounts of muscle of each side (Fig. 5.7).

In rectoperineal fistula, the incision circumscribes the anus anteriorly and extends to the tip of the coccyx posteriorly. The fistula is mobilised with the assistance of multiple stay sutures in the mucocutaneous junction. Once adequate mobilisation has been achieved to place the rectum within the centre of the sphincter complex without tension, the rectum is anchored to the muscle complex posteriorly and the anoplasty completed.

In rectovestibular fistula in girls, the incision is similar but longer to achieve adequate mobilisation. The incision is extended anteriorly remaining in the midline to the posterior fourchette of the vaginal introitus and around the opening of the fistula in the shape of a tennis racket. The most important component of this particular operation is the requirement to completely separate the rectum from the vagina as the two structures are very adherent. A plane of dissection is created proceeding cranially until each viscus separates and has its own full thickness wall. This meticulous dissection is best performed with fine-needle cautery. Once the rectum has been adequately mobilised, it is positioned within the centre of the muscle complex. Anteriorly the perineal body is reconstructed. The anoplasty is completed by securing the rectum to the skin.

For repair of a rectourethral fistula, a longer incision and deeper dissection are required. The incision remains precisely in the midline having first identified the location of the muscle complex

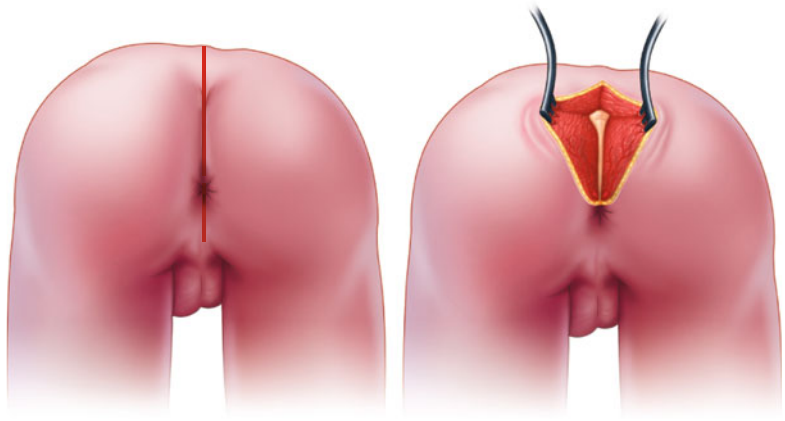

Fig. 5.7 Posterior Sagittal Anorectoplasty (PSARP) showing midline incision and deeper dissection through fat and muscle complex (Figure 27.7, page 295 from *Pediatric Surgery* (Springer Surgery Atlas Series), Eds: Puri & Hollwarthy, Springer 2006: ISBN 978-3-540-40738-3)

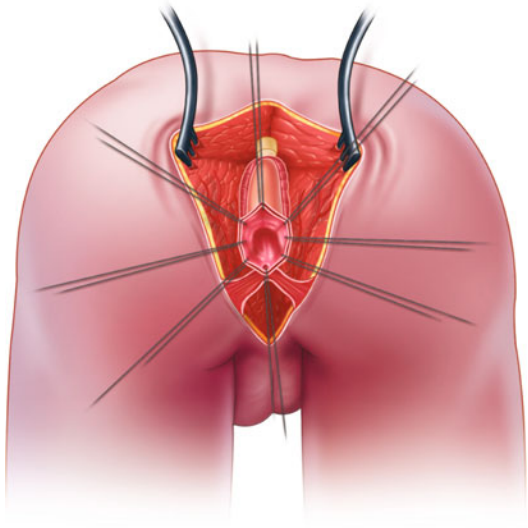

Fig. 5.8 Identification of the fistula from within the rectal lumen in cases of rectourethral fistula (Figure 27.5a-c, page 293 from *Pediatric Surgery* (Springer Surgery Atlas Series), Eds: Puri & Hollwarthy, Springer 2006: ISBN 978-3-540-40738-3)

with electrical stimulation. Our practice is to mark the skin overlying the centre of the muscle complex on either side of the midline to facilitate later correct placement of the neorectum. Having divided the muscle complex, the dissection continues in the midline, proceeding deeper through a layer of fat (the ischiorectal space) to the levator muscle whose fibres run medial and perpendicular to the parasagittal fibres of the muscle complex. The dissection continues in the midline until the rectum is encountered. The rectum is opened longitudinally in the midline between stay sutures and the fistula site identified from within the rectal lumen (Fig. 5.8). The rectum and the urethra share a common wall just proximal to the fistula opening. A submucosal plane must be dissected for at least 1 cm proximal to the fistula following which the dissection continues in a plane taking the full thickness of the rectal wall. Once the rectum has been completely separated from the urethra, the fistula is closed with interrupted absorbable sutures. The dissection continues around the circumference of the rectum until adequate mobilisation has been achieved to bring the rectum to the perineal skin without tension. Further length is obtained by division of blood vessels on the wall of the rectum. The rectum is placed anterior to the muscle of levator ani and within the muscle complex. Finally the anoplasty is completed suturing the rectal end of the fistula to the skin.

For very high defects including all recto-bladder neck fistulae and some rectoprostatic fistulae in males, the perineal approach must be combined with an abdominal approach. Traditionally a laparotomy was required to identify and divide such high fistulae and achieve adequate rectal mobilisation to perform the reconstruction. However, with the advent of minimally invasive surgery, the abdominal part of the operation is now performed laparoscopically. Some surgeons have reported a modification to the formal posterior sagittal approach to anorectoplasty for this type of anomaly involving blunt dissection through the muscle complex up into the pelvis. This obviates the need to formally divide the muscles as in the formal PSARP. Children with such a high anomaly typically have very poor pelvic muscles and rarely achieve good bowel control regardless of the operation employed. Hence, although the long-term results of this change in technique are not clear, we favour this altered approach.

With the infant in a supine position, pneumoperitoneum is created using a supra-umbilical open approach and two further trocars are introduced under vision. The fistula is identified by following the sigmoid colon into the pelvis having opened the peritoneal reflection. The terminal part of the colon enters the fistula which in turn enters the bladder neck or prostatic urethra. The orientation of the fistula in these cases is usually in a T-shaped arrangement without a long common wall. The fistula is divided and ligated as close as possible to the urinary tract. The colon is then mobilised to achieve enough length to reach the perineum. This may be difficult and particular attention must be paid to the rectal blood supply which is predominantly intramural. Following adequate mobilisation attention is turned to the perineum. Having identified the location of the sphincter complex with electrostimulation, a tunnel is developed from the perineum below, the bowel brought down through this and sutured to skin.

Posterior Sagittal Anorectovaginourethroplasty (PSARVUP)

The posterior sagittal approach is also used to repair cloacal malformations. These anomalies form part of a wide spectrum of structural disorders with a common channel of variable length. As the anatomy is very variable and inconsistent, the procedure is often performed by a paediatric surgeon in conjunction with a paediatric urologist. The operative technique of total urogenital mobilisation is most commonly employed. The urethra, vagina and bowel are mobilised en bloc before separating the different structures. This technique has greatly improved outcome and in particular the avoidance of urethrovaginal fistulae and vaginal stenosis. In infants where the common channel is very long (>3 cm), reconstruction may be very difficult. It may be close to impossible to reconstruct an adequate urethra and vagina. In some, vaginal hypoplasia or absence may mandate some form of vaginal substitution. This may necessitate staged reconstruction performed at an older age.

Post-operatively infants with a low malformation who have undergone limited PSARP may be discharged once they are passing stool regularly and all diagnostic imaging has been performed. Following repair of a higher anomaly, a urethral catheter should remain in situ for a minimum of a week. Catheter removal is done in hospital and discharge home occurs when adequate voiding is established.

Wound infection and dehiscence remain a real concern; intravenous antibiotics are continued for 5–7 days and the infants are nursed prone to keep pressure off the wound. Some surgeons recommend routine examination under anaesthetic and anal calibration 2 weeks following the procedure. The use of regular anal dilatation by the parents for several months is popular and widely recommended. In our experience this is distressing for parents and child and is unnecessary. We prefer to perform an examination under anaesthesia several months following anoplasty prior to stoma closure. If the anus is of satisfactory calibre, the stoma can be closed.

Outcome

Outcome is related to a number of different factors including the anatomy of the original malformation and degree of associated anomalies, in particular spinal or sacral problems. As a general principle, higher anomalies are associated with a worse long-term functional outcome as are those with an associated sacral or spinal abnormality. It should be remembered that the aim of anorectal reconstructive surgery is to achieve a normal functional outcome. This includes being able to pass stool voluntarily without soiling or significant constipation. Overall, in large series of patients comprising the full spectrum of anorectal anomalies, voluntary bowel motions are reported by 70 % of patients, but only about half reports total continence. The remainder reports regular soiling and/or significant constipation. Whilst ‘normal’ bowel function can be achieved in up to 80–90 % of patients with a rectoperineal fistula, only 10–15 % of patients with a rectobladder neck fistula experience such a good outcome.

An important consideration is that what is generally considered to be normal stooling function is in fact not achieved by 100 % of healthy individuals. A recent study of healthy Finnish patients reported normal stooling frequency in only 92 % of adults and that 10 % of adults reported some degree of social compromise as a result of disordered bowel function. Just over one-quarter of adults reported faecal soiling with a frequency of less than once per week and 3.5 % reported faecal accidents with an identical frequency. These findings have implications for the assessment of outcome of children born with anorectal anomalies. In attempting to achieve ‘normality’, it is important to consider what normal bowel function actually is.

For children and their families, the consequences of an abnormal stooling pattern and in particular frequent faecal soiling are significant. These children are best treated with a bowel management programme in order to achieve ‘social continence’ to enable the child to come out of nappies and attend school without disruption. A programme of daily or alternate day

enemas is commenced to completely evacuate the colon and avoid faecal incontinence during the following 24–48 h. By manipulating this regime of enemas and combining it with an oral laxative preparation, incontinence and soiling can be 'managed' in many children such that the impact on daily life can be minimised and child and family quality of life improved. Some children and their families do not tolerate rectal enemas well and may benefit from surgical creation of an antegrade continence enema (ACE) channel. This involves surgical creation of an appendicostomy to allow antegrade instillation of an enema solution. Such a procedure is usually not tolerated below the age of 7 years due to lack of motivation and cooperation from the child.

Long-term outcomes into adolescence and beyond are only recently being reported for children born with anorectal malformations. Encouragingly the majority of children experience some improvement around the time of puberty. The outcome in regard to sexual function and fertility is not clear at the present time. Our experience, which predates use of the PSARVUP technique, suggests that pregnancy was unusual in girls with high anomalies.

Further Reading

Bischoff A, Levitt MA, Peña A. Laparoscopy and its use in the repair of anorectal malformations. J Pediatr Surg. 2011;46(8):1609–17.

Georgeson K. Laparoscopic-assisted anorectal pull-through. Semin Pediatr Surg. 2007;16(4):266–9.

Herman RS, Teitelbaum DH. Anorectal malformations. Clin Perinatol. 2012;39(2):403–22.

Kluth D, Hillen M, Lambrecht W. The principles of normal and abnormal hindgut development. J Pediatr Surg. 1995;30(8):1143–7.

Kyrklund K, Koivusalo A, Rintala RJ, Pakarinen MP. Evaluation of bowel function and fecal continence in 594 Finnish individuals aged 4 to 26 years. Dis Colon Rectum. 2012;55(6):671–6.

Levitt MA, Bischoff A, Peña A. Pitfalls and challenges of cloaca repair: how to reduce the need for reoperations. J Pediatr Surg. 2011;46(6):1250–5.

Rintala RJ, Pakarinen MP. Imperforate anus: long- and short-term outcome. Semin Pediatr Surg. 2008; 17(2):79–89.

Sen G, Sebire NJ, Olsen O, Kiely E, Levitt GA. Familial Currarino syndrome presenting with peripheral primitive neuroectodermal tumour arising with a sacral teratoma. Pediatr Blood Cancer. 2008;50(1):1–4.

Van der Putte SC, Neeteson FA. The pathogenesis of hereditary congenital malformations of the anorectum in the pig. Acta Morphol Neerl Scand. 1984;22(1): 17–40.

Warne SA, Hiorns MP, Curry J, Mushtaq I. Understanding cloacal anomalies. Arch Dis Child. 2011;96(11): 1072–6.

Premalignant Conditions of the Anus

6

James Crosbie, Jonathan McCullough, and Paul Boulos

Introduction

The diagnosis of premalignant conditions of the anus remains a challenge, in particular differentiating between lesions which have a malignant potential and those which are of no concern. The use of anal cytology and high-resolution anoscopy (HRA) has helped to diagnose clinically suspicious lesions. In addition, HRA allows biopsy and treatment of clinically suspicious lesions. These investigations can be used to screen high-risk patients, such as HIV-positive men who have sex with men. The human papillomavirus is implicated in the aetiology of many premalignant lesions, and vaccination against it in at-risk populations may reduce the prevalence of these lesions.

The premalignant conditions of the anus which will be discussed in this chapter are:

1. Anal intraepithelial neoplasia (AIN)
2. Paget's disease of the anus
3. Bowen's disease of the anus

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Clinical Assessment

It is important to ascertain if the patient has any risk factors for developing premalignant conditions of the anus such as HIV status, sexual history, history of immunotherapy and history of prior anal, perianal or gynaecological lesions. Examination of patients involves thorough inspection of the perianal skin and anal margin, a digital rectal examination and proctoscopy. At-risk patients should have anal cytology. Patients with abnormal cytology or lesions identified on initial examination should be referred for high-resolution anoscopy allowing biopsy and/or treatment of suspicious lesions.

Symptoms

Patients are often asymptomatic; however, common symptoms are pruritus ani and bleeding. The patient may notice an anal lesion, though often worrying lesions are flat.

Signs

On anal inspection, a premalignant lesion may present as a plaque. AIN may be found in anal warts. AIN lesions may be eczematoid, papillomatous, papular or plaque-like. Induration and ulceration are more sinister signs of invasion.

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Anal Cytology

Anal cytology involves rotating a water-moistened Dacron swab inside the anal canal. Dacron is used instead of cotton because cells cling to cotton and this may decrease the cellular yield. To increase the cellular exfoliative yield, patients are instructed to refrain from anal sex and avoid douching or having enemas before testing. The cytological findings are classified as:

1. Normal
2. Atypical squamous cells of undetermined significance (ASCUS)
3. Low-grade squamous intraepithelial lesion (LSIL)
4. High-grade squamous intraepithelial lesions (HSIL)

The sensitivity of anal cytology is 90 % but its specificity is 50 %. It is suggested that all patients with abnormal cytology are referred for HRA and biopsy of visible lesions. While HSIL has a high predictive value for detection of biopsy-proven AIN 2 or 3, especially in HIV-positive patients, ASCUS or LSIL are not reliable for the determination of the true grade of the lesion [1].

High-Resolution Anoscopy

High-resolution anoscopy is similar to cervical colposcopy and uses identical equipment (a powerful light source and a binocular lens). Lesions identified on HRA are characterised by colour, contour, surface configuration and vascular patterns.

Colour

Acetic acid is applied to the mucosa, and 'acetowhitening' of the abnormal tissue compared with surrounding normal tissue is observed, which can be improved by the administration of Lugol iodine solution, in which the abnormal cells remain unstained or appear yellow, in contrast to the mahogany colour of the surrounding stained normal tissue.

Contour

This is described as regular or irregular.

Surface Configuration

Lesions are described based on the presence or absence of papillae, raised or flat, smooth or granular and papillary or non-papillary.

Vascular Patterns

The vascular pattern may be normal or warty. In addition vascular punctuation, which is seen as a series of dots on acetowhite epithelium and represents the end on view of dilated and prominent intraepithelial capillaries, can be coarse or fine.

Mosaicism is caused by normal interconnection of capillaries causing a tile-like pattern and it can be coarse or fine.

Anal Intraepithelial Neoplasia

Anal intraepithelial neoplasia (AIN) was first described in 1986 by Fenger and Neilsen [2]. AIN is a multifocal premalignant disease process that affects the perianal skin and the anal canal including the anal transition zone. It has three grades (Fig. 6.1):

AIN 1: nuclear abnormalities affect the lower one-third of the epithelium.

AIN 2: nuclear abnormalities affect the lower two-thirds of the epithelium.

AIN 3: nuclear abnormalities involving the full thickness of the epithelium.

The term anal squamous intraepithelial lesions is also used to describe AIN and is classified into low-grade anal squamous intraepithelial lesions (LSIL) which is equivalent to AIN grade 1 and high-grade anal squamous intraepithelial lesions (HSIL) which is equivalent to AIN grade 2 or 3.

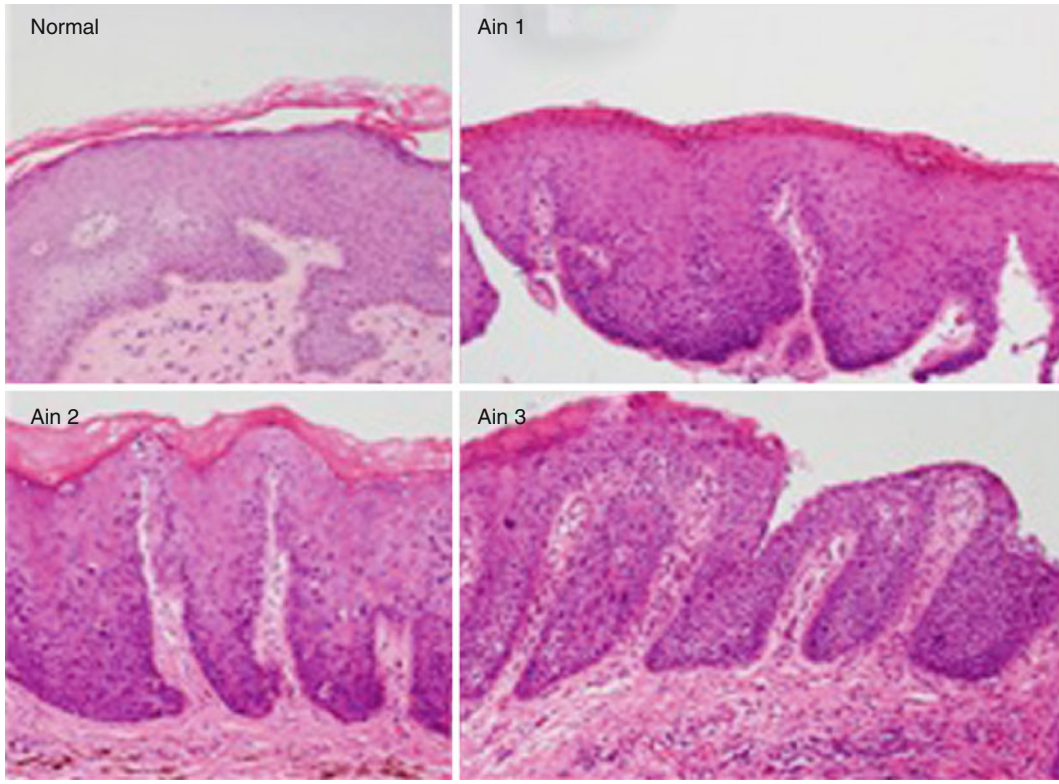

Fig. 6.1 AIN lesions

AIN is more common in the immune-suppressed patient and in particular in HIV-positive patients. The prevalence of AIN in HIV-positive men who have sex with men (MSM) is estimated to be around 26–36 %. The prevalence of high-grade AIN in this subgroup is around 5 % [3, 4].

AIN is associated with the human papillomavirus (HPV), which is usually transmitted by genital contact during sexual intercourse [5]. The risk of persistence and progression of the HPV infection to precancerous lesions varies by the HPV type [6]. There are over 100 HPV types identified and about 40 of these can cause anogenital disease in humans [7]. They are graded according to their association with cervical cancer into low-, intermediate- and high-risk subgroups:

Low-risk HPV Types	*6*, *11*, 40, *42*, *43*, *44*, 54, 61, 70, 72
Medium-risk HPV Types	26, 53, 66
High-risk HPV Types	*16*, *18*, *31*, 33, 35, 39, *45*, 51, 52, 56, 58, 59, 68, 73, 82

HPV types 16 and 18 are most commonly isolated in cervical, anal, vulvar and penile cancer. Among men who have sex with men (MSM) and women (commercial sex workers and intravenous drug users) who are HIV positive, the incidence of HPV infection, multiplicity of HPV types and abnormal anal or cervical cytology and the prevalence of AIN are higher than in HIV-negative men and women [8]. The progression from low- to high-grade AIN is related to HIV status and occurs in more than 50 % of

HIV-positive patients within 2 years [9]. The rate of progression to squamous cell carcinoma is probably <1 % per annum [9]. It is thought that HPV infections occur in areas of trauma and result in cellular hyperplasia of the basal layer of the squamous, transitional and cuboidal epithelial cells. Viral genomes have been cloned from infected tissue. HPV E6 and E7gene products inactivate p53 and Rb protein products of tumour suppressor genes. This is postulated to result in inactivation of their tumour suppressor function under appropriate circumstances, such as deficient immune state, thereby allowing malignant transformation and tumorigenesis [10, 11].

Diagnosis of AIN

HPV-associated premalignant conditions are often asymptomatic. The diagnosis of AIN should always be considered in high-risk patients who include all men who have sex with men (MSM) regardless of HIV infection; women with cervical intraepithelial neoplasia (CIN), high-grade vulva disease or cancer; all HIV-positive men and women regardless of sexual orientation; individuals with perianal condylomata; and transplant recipients. Kreuter et al. have classified commonly seen lesions into bowenoid, erythroplakia and verrucous [12]. Examination of the external genitalia should be performed in both sexes and women should be offered colposcopy. As AIN is often multifocal, patients should have mapping biopsies performed. Mapping involves performing four quadrant biopsies of the anal canal, anal margin and perianal skin and also biopsying any suspicious lesions. Scholefield et al. first described the use of high-resolution anoscopy with acetowhitening of the anal mucosa in diagnosing AIN [13]. Subsequently Palefsky described in detail the colposcopic appearances of anal squamous intraepithelial lesions [14]. High-grade AIN is usually smooth and flat with vascular punctuation. It is uncommon for high-grade AIN to have papillae, warty vessels or mosaicism. However, it is advised that any abnormal lesion is biopsied to confirm its histology.

Screening for AIN

A protocol for AIN screening was proposed by Palefsky et al. [1]. The first step involved is taking anal cytology followed by confirmation of the disease stage by high-resolution anoscopy and biopsy. HRA testing has a high predictive value for normal mucosa and for AIN 3 but not for AIN 1 or 2 [14]. Patients with normal cytology or normal HRA are rescreened in 1 year if they are HIV positive or 2–3 years if HIV negative. Patients with AIN 1 are screened every 6 months. AIN 2 and 3 will require treatment.

Treatment of AIN

The rationale for treating AIN is to prevent the progression into high-grade and invasive cancer. Sceptics argue that involvement of AIN 2 or 3 of deeper areas of the perianal skin and its appendages in up to 50 % of cases and sweat glands in 25 % of biopsy specimens may not be accessible to anal colposcopy, and ablative techniques such as cryotherapy, laser vaporisation and electrocautery are inadequate forms of treatment [15]. HPV is epitheliotropic, the entire genital area is likely to be involved, and eliminating the virus may not be possible. AIN 1 and 2 could be treated expectantly since their natural course is unclear and occasionally regress spontaneously; furthermore, they are frequently associated with HPV 6 and 11 which is less oncogenic than HPV 16 [16].

Small perianal lesions are treated by topical agents that include podophyllotoxin, Imiquimod, Cidofovir, 80 % trichloroacetic acid and liquid nitrogen. Small intra-anal lesions can be treated with 80 % trichloroacetic acid. Medium and larger lesions are treated by surgical excision, electrocautery, laser ablation, photodynamic therapy or infrared coagulation. The advantage of surgical excision is that the tissue can be sent for histological analysis. Regardless of the mode of treatment, local recurrence is common. Chang et al. reported local recurrence in two-thirds of patients treated with HRA-directed surgical excision of intra-anal lesions [17]. Goldstone et al. reported a similar recurrence rate for treating

AIN using infrared coagulation under local anaesthesia [18]. Ablative techniques must ensure a depth of penetration 2.2 mm below the basement membrane to adequately treat the disease.

Imiquimod Cream

Imiquimod cream as a treatment option has the advantage of being applied by the patient and also it can be used for extensive disease. It is an immune stimulant with minimal side effects. Systemic absorption can cause transient flulike symptoms the day after application. Imiquimod cream has been shown to be effective in treating vulva intraepithelial neoplasia. Van Seters et al. demonstrated a 35 % clearance rate of VIN 2/3 after a 12-month treatment period and an 81 % improvement in symptoms [19]. Imiquimod cream is not licensed for use in or around the anus; however, initial studies have shown considerable promise. In an uncontrolled study by Wieland et al., 77 % of lesions resolved after 16 weeks of treatment and were associated with a reduction of HPV viral load. However, approximately half of the patients in the study had low-grade AIN [20]. More recently Fox et al., in a double-blind randomised placebo-controlled trial, demonstrated that Imiquimod cream has a role in treating high-grade AIN. It can be used as an adjuvant to or alternating with other treatment for high-grade multifocal AIN. High-grade AIN resolved in 4 out of 28 patients and was downgraded in another 8 patients [21].

Cidofovir Cream

A 1 % gel used topically for 5 days a week for up to 6 weeks has been shown to be more effective than electrosurgery in the treatment of anogenital warts in HIV-positive patients [22]

Electrocautery

Treating lesions in the anal canal with electrocautery has been disappointing with a high recurrence rate (79 % in 1 year and 100 % in 50 months) [17].

Infrared Coagulation

This procedure is better tolerated than electrocautery. Goldstone pioneered this technique and showed an initial response rate of 72 % per treatment cycle [18]. Infrared coagulation can be performed in the outpatient setting with administration of local anaesthetic. It provides good haemostasis and no smoke plume. It uses an infrared beam of light pulsed at 0.5–3.0 s intervals and prevents damage to the deeper tissues. The depth of penetration in millimetres is approximately equal to the length of pulse applied in seconds. For example, a 1 s pulse penetrates approximately 1 mm of tissue. Typically a lesion is treated by touching it with IRC and firing for 1.5 s and repeating as necessary to remove the lesion. Patients can expect 1–2 weeks of pain and bleeding with bowel movements.

Photodiagnosis and Photodynamic Therapy for AIN

There are a few studies showing the feasibility of photodynamic therapy in treating AIN. 5-Aminolevulinc acid (ALA), when added to many tissues, results in the accumulation of sufficient quantities, especially in tumour and dysplastic cells, of the endogenous photosensitizer protoporphyrin IX, which exhibits red fluorescence with blue light and produces a photodynamic effect when exposed to activating light by generating cytotoxic singlet oxygen. Topical application of ALA followed by exposure to activating light has been used as a treatment for AIN.

Human Papillomavirus Vaccination

HPV vaccination is safe and well tolerated. There are two vaccines available:

1. Gardasil—a quadrivalent vaccine which protects against HPV 6, 11, 16 and 18
2. Cervarix—a bivalent vaccine which protects against HPV types 16 and 18

Both vaccines result in a decrease in anogenital abnormalities [23, 24].

However, these vaccines do not exert therapeutic effects on existing lesions and are unlikely to have an immediate impact on the prevalence of anogenital cancer and its precursor lesions. It is suggested that therapeutic HPV vaccines could result in the elimination of pre-existing lesions and infections by generating cellular immunity against HPV-infected cells. These vaccines could play a major role in controlling HPV-associated premalignant lesions in the future [25].

Bowen's Disease of the Anus

Bowen's disease was first described by John T. Bowen in 1912 [26]. It is an intraepithelial squamous cell carcinoma of the skin. While it is more common on the trunk, it can also occur in the perianal region and perineum where it is associated with the human papillomavirus types 16 and 18 [27–30]. Bowen's disease is more likely to present in female patients than males and it commonly presents in the fifth decade [30, 31]. In comparison with extramammary Paget's disease, the incidence of internal malignancy in Bowen's disease is low (5 vs. 52–73 %), and the incidence of invasive carcinoma at the site of the lesion is also low (2–6 vs. 50 %). Bowen's disease is often asymptomatic and found coincidentally in haemorrhoidectomy specimens (25–40 %). Often there is a delay in diagnosis of months to years due to its slow-growing nature and the paucity of symptoms [31, 32]. Patients who do develop symptoms tend to complain of pruritus, perianal bleeding and anal discharge. On clinical examination, there may be a dry scaly raised lesion with an irregular margin and brownish-red eczematoid appearance [27, 30, 33]. Microscopically Bowen's disease is characterised by hyperchromatic enlarged nuclei with easily identifiable mitotic figures, parakeratosis and hyperkeratosis in the superficial surface layers, loss of cellular polarity, marked epidermal hyperplasia with elongation and thickening of the rete ridges and randomly distributed dyskeratotic cells with densely prematurely enlarged eosinophilic cytoplasm [15]. Large haloed hyperchromatic nuclei are present and stain negative with periodic acid-Schiff stain for glycogen [27, 31]. This is in contrast to Paget's disease which stains for carcinoembryonic antigen and usually mucin [30, 33]. It is thought that the natural history of Bowen's disease is relatively benign with an estimated 2–6 % conversion to invasive squamous cell carcinoma [28, 31, 34]. The treatment options available for Bowen's disease are similar to those for AIN and include wide local excision with primary closure [35, 36], wide local excision with split-thickness skin graft [37, 38], wide local excision with advancement cutaneous anoplasty [30, 39, 40], local excision of macroscopic lesions alone [35, 36], cautery fulguration [10], CO_2 laser ablation [31, 41], cryotherapy [10, 42, 43], topical 5-fluorouracil cream [41], photodynamic therapy [44], argon beam laser therapy [41, 45] and observation alone [15].

Paget's Disease of the Anus

In 1874 Sir James Paget described 15 patients in whom the nipple and the surrounding skin of the breast was diseased, and within 2 years all patients developed an ipsilateral breast carcinoma. Extramammary Paget's disease (EMPD) was first described by Crocker in 1889 [46], and in 1893 Darier and Coulillaud described EMPD of the anogenital area.

Extramammary Paget's disease (EMPD) can be classified as primary or secondary. Primary EMPD is an in situ adenocarcinoma of apocrine gland ducts or pluripotent keratinocyte stem cells [47]. Secondary EMPD is an intraepithelial spread due to an underlying carcinoma of the skin or other organ systems. EMPD affects areas of the skin containing apocrine sweat glands with approximately 65 % affecting the vulva, 15 % the perianal area, and 14 % the penis, scrotum and inguinal regions. EMPD is a rare neoplasm with only a few hundred cases reported in the literature. EMPD occurs in older patients and is more common in women than men. The lesion spreads epidermotropically for years and even decades prior to becoming invasive. The risk of an associated cancer is high and all patients should be

thoroughly investigated. It is thought that there is a 25–35 % risk of an associated cancer in perianal extramammary Paget's disease [48].

The commonest symptom in perianal EMPD is intense pruritus, while pain and bleeding may occur later. Most patients present with a non-resolving eczematous lesion of the perineum. In the early stages, the clinical signs are often subtle, and they include some erythema, skin excoriation and lichenification. In an established EMPD lesion, there will be a sharply defined erythematous plaque of abnormal tissue which may have superficial erosions. The differential diagnosis includes Bowen's disease, basal cell carcinoma, dermatitis, cutaneous candidiasis and intertrigo.

The diagnosis of EMPD is usually made by histological analysis which shows Paget cells. These are large, faintly basophilic or vacuolated cells situated at the dermoepidermal junction. The nuclei are vesicular and either central or compressed at the periphery. Paget cells contain a lot of mucin and carcinoembryonic antigen [49]. There is little mitotic activity. They also express gross cystic disease fluid protein, which can be regarded as a specific marker for that cell [50]. Gross cystic disease fluid protein (GCDFP-15) is a marker of apocrine glands and is strongly expressed in perianal EMPD. It is thought that expression of the c-ErbB-2 oncoprotein may play a role in promoting intraepithelial spread of adenocarcinoma cells [51]. There is also a variable degree of hyperkeratosis and acanthosis. The superficial dermis shows a chronic inflammatory infiltrate with vascular dilatation and engorgement.

Following confirmation of the diagnosis, all patients should have a full skin examination, palpable of all lymph nodes, rectal examination, sigmoidoscopy and cystoscopy. In addition female patients should have a full gynaecological assessment to include breast examination and colposcopy.

Treatment of EMPD is predominantly surgical; however, there have been some case reports detailing the use of chemotherapeutic agents 5-fluorouracil [52], Imiquimod [52] and a combination of paclitaxel and trastuzumab [53]. Topical agents may have a role when surgery is contraindicated in high-risk patients, and its future will need to be confirmed by more studies [54].

The treatment of primary EMPD is wide local excision of the affected area with a margin of histologically normal skin [49]. However, traditional surgical excision is associated with high recurrence rates (30–60 %). Mohs micrographic surgery is associated with a lower rate of recurrence (8–26 %) and a smaller margin of normal skin excised with the specimen [55, 56]. The average time to recurrence is 2.5 years. Mohs micrographic surgery involves removing the surgical specimen, analysing the tissue and its margins and re-excising any involved margin until such time as all margins are clear. It was first described by Dr. Frederic Mohs in 1938 while he was still a medical student. Follow-up of patients after surgery should be every 3 months for 2 years and annually after that if there are no signs of recurrence. Treatment of secondary EMPD involves mainly treating the underlying cancer.

Summary

Premalignant lesions of the anus remain a challenge, in relation to both diagnosis and subsequent treatment. Traditional surgical treatment of wide local excision has been associated with a high recurrence rate and significant morbidity. Screening of high-risk patients with anal cytology and subsequent high-resolution anoscopy-directed treatment of early lesions has soon considerable promise. Prevention of premalignant lesions of the anus may be achieved by vaccination of patients in their teens against the human papillomavirus.

References

1. Palefsky JM, Holly E, Hogeboom CJ. Anal cytology as a screening tool for anal squamous intraepithelial lesions. J Acquir Immune Defic Syndr Hum Retrovirol. 1997;14:415–22.
2. Fenger C, Neilsen VT. Intraepithelial neoplasia in the anal canal. Acta Pathol Microbiol Immunol Scand A. 1986;94:343–9.

3. Palefsky JM, Holly EA, Ralston ML. Anal squamous intraepithelial lesions in HIV positive and HIV negative homosexual and bisexual men. J Acquir Immune Defic Syndr Hum Retrovirol. 1998;17:320–6.
4. Kiviat NB, Critchlow CW, Holmes KK. Association of anal dysplasia and HPV with immunosuppression and HIV infection among homosexual men. AIDS. 1993;7:43–9.
5. Winer RL, Lee SK, Hughes JP. Genital human papillomavirus infection: incidence and risk factors in a cohort of female university students. Am J Epidemiol. 2003;157:218–26.
6. Wheeler CM, Hunt WC, Schiffman M. Human papillomavirus genotypes and the cumulative 2-year risk of cervical precancer. J Infect Dis. 2006;194:1291–9.
7. Palefsky J, Holly EA. Molecular virology and epidemiology of human papilloma virus and cervical cancer. Cancer Epidemiol Biomarkers Prev. 1995;4:451–8.
8. Chin-Hong PV, Palefsky JM. Natural history and clinical management of anal human papillomavirus disease in men and women infected with human immunodeficiency virus. Clin Infect Dis. 2002;35(9):1127–34.
9. Klencke BJ, Palefsky JM. Anal cancer: an HIV associated cancer. Hematol Oncol Clin North Am. 2003; 17:850–72.
10. Northfelt DW, Swift PS, Palefsky JM. Anal neoplasia. Pathogenesis, diagnosis, and management. Hematol Oncol Clin North Am. 1996;10:1177–87.
11. Surawicz CM, Kiviat NB. A rational approach to anal intraepithelial neoplasia. Semin Colon Rectal Surg. 1998;9:99–106.
12. Kreuter A, Brockmeyer NH, Hochdorfer B, Weissenborn SJ, Stucker M, Swoboda J, et al. Clinical spectrum and virologic characteristics of anal intraepithelial neoplasia in HIV infection. J Am Acad Dermatol. 2005;52(4):603–8.
13. Scholefield JH, Sonnex C, Talbot IC, Palmer JG, Whatrup C, Mindel A, et al. Anal and cervical intraepithelial neoplasia: possible parallel. Lancet. 1989; 2(8666):765–9.
14. Jay N, Berry JM, Hogeboom CJ. Colposcopic appearance of anal squamous intraepithelial lesions: relationship to histopathology. Dis Colon Rectum. 1987;40:919–28.
15. Cleary RK, Schaldenbrand JD, Fowler JJ, Schuler JM, Lampman RM. Perianal Bowen's disease and anal intraepithelial neoplasia: review of the literature. Dis Colon Rectum. 1999;42(7):945–51.
16. Zbar AP, Fenger C, Efron J, Beer-Gabel M, Wexner SD. The pathology and molecular biology of anal intraepithelial neoplasia: comparisons with cervical and vulvar intraepithelial carcinoma. Int J Colorectal Dis. 2002;17(4):203–15.
17. Chang GC, Berry JM, Jay N. Surgical treatment of high-grade anal squamous intraepithelial lesions: a prospective study. Dis Colon Rectum. 2002;45:453–8.
18. Goldstone SE, Zawalek MD, Huyett JW. Infrared coagulation: a useful tool for treating anal squamous intraepithelial lesions. Dis Colon Rectum. 2005;48:1042–54.
19. van Seters M, van Beurden M, Kate JW, Eijkemans M, Kagie M. Treatment of vulvar intraepithelial neoplasia with topical imiquimod. N Engl J Med. 2008;358:1465–73.
20. Wieland U, Brockmeyer NH, Weissenborn SJ, Hochdorfer B, Stucker M, Swoboda J, et al. Imiquimod treatment of anal intraepithelial neoplasia in HIV-positive men. Arch Dermatol. 2006;142(11): 1438–44.
21. Fox PA, Nathan M, Francis N, Singh N, Weir J, Dixon G, et al. A double-blind, randomized controlled trial of the use of imiquimod cream for the treatment of anal canal high-grade anal intraepithelial neoplasia in HIV-positive MSM on HAART, with long-term follow-up data including the use of open-label imiquimod. AIDS. 2010;24(15):2331–5.
22. Orlando G, Fasolo MW, Beretta R. Combined surgery and cidofovir is an effective treatment for genital warts in HIV-infected patients. AIDS. 2002;16(3): 447–50.
23. Paavonen J, Lehtinen M. Introducing human papillomavirus vaccines-questions remain. Ann Med. 2008;40:162–6.
24. Rogers LJ, Eva LJ, uesley DM. Vaccines against cervical cancer. Curr Opin Oncol. 2008;20:570–4.
25. Ma B, Xu Y, Hung C. HPV and therapeutic vaccines: where are we in 2010? Curr Cancer Ther Rev. 2010;6:81–103.
26. Bowen JT. Precancerous dermatoses: a study of 2 cases of chronic atypical epithelial proliferation. J Cutan Dis. 1912;30:241–55.
27. Beck DE. Paget's disease and Bowen's disease of the anus. Semin Colon Rectal Surg. 1995;6:143–9.
28. Stearns Jr MW, Grodsky L, Harrison Jr EG, Quan S, Rob CG. Malignant anal lesions. Dis Colon Rectum. 1966;9:315–27.
29. Marfing TE, ABel ME, Gallagher DM. Perianal Bowen's disease and associated malignancies: results of a survey. Dis Colon Rectum. 1987;30:782–5.
30. Sarmiento JM, Wolff BG, Burgart LJ, Frizelle FA, Ilstrup DM. Perianal Bowen's disease: associated tumours, human papillomavirus, surgery and other controversies. Dis Colon Rectum. 1997;40: 912–8.
31. Marchesa P, Fazio VW, Oliart S, Goldblum JR, Lavery IC. Perianal Bowen's disease: a clinicopathologic study of 47 patients. Dis Colon Rectum. 1997;40:1286–93.
32. Beck DE, Fazio VW. Perianal Bowen's disease. Dis Colon Rectum. 1988;31:419–22.
33. Beck DE, Fazio VW. Premalignant lesions of the anal margin. South Med J. 1989;82:470–4.
34. Schwartz RA, Stoll Jr HL. Epithelial precancerous lesions. New York: McGraw-Hill; 1993. p. 804–21.
35. Ramos R, Salinas H, Tucker L. Conservative approach to the treatment of Bowen's disease of the anus. Dis Colon Rectum. 1983;26:712–5.
36. Rasmussen OO, Christiansen J. Conservative management of Bowen's disease of the anus. Int J Colorectal Dis. 1989;4:164–6.

37. Seckel BR, Schoetz Jr DJ, Coller JA. Skin grafts for circumferential coverage of perianal wounds. Surg Clin North Am. 1985;65:365–71.
38. Reynolds VH, Madden JJ, Franklin JD, Burnett LS, Jones HW. Preservation of anal function after total excision of the anal mucosa for Bowen's disease. Ann Surg. 1984;199:563–8.
39. Core GB, Bite U, Pemberton JH, Petty PM. Sliding V-Y perineal island flaps for large perianal defects. Ann Plast Surg. 1994;32:328–31.
40. Sentovich SM, Falk PM, Christensen MA, Thorson AG, Blatchford GJ, Pitsch RM. Operative results of house advancement anoplasty. Br J Surg. 1996;83: 1242–4.
41. Bandieramonte G, Bono A, Zurrida S, Bartoli C, de Palo G. Laser surgery for small perianal neoplasms. Eur J Cancer. 1993;29:1528–31.
42. Morton CA, WHitehurst C, Moseley H, McColl JH, Moore JV, Mackie RM. Comparison of photodynamic therapy with cryotherapy in the treatment of Bowen's disease. Br J Dermatol. 1996;135:766–71.
43. Holt PJ. Cryotherapy for skin cancer: results over a 5-year period using liquid nitrogen spray cryosurgery. Br J Dermatol. 1988;119:231–40.
44. Petrelli NJ, Cebollero JA, Rodriquez-Bigas M, Mang T. Photodynamic therapy in the management of neoplasms of the perianal skin. Arch Surg. 1992;127:1436–8.
45. Boynton KK, Bjorkman DJ. Argon beam laser for perianal Bowen's disease: a case report. Lasers Surg Med. 1991;11:385–7.
46. Crocker HR. Paget's disease affecting the scrotum and penis. Trans Pathol Soc Lond. 1889;40:187–91.
47. Lloyd J, Flanagan AM. Mammary and extramammary Paget's disease. J Clin Pathol. 2000;53:742–9.
48. Goldblum JR, Hart WR. Perianal Paget's disease – a histologic and immunohistochemical study of 11 cases with and without associated rectal adenocarcinoma. Am J Surg Pathol. 1998;2:170–9.
49. Beck DE, Fazio VW. Perianal Paget's disease. Dis Colon Rectum. 1987;30:263–6.
50. Messinetti S, Giacomelli L, Finizio R. Perinal Paget's disease: clinical observation and review of the literature. Ann Ital Chir. 1994;65:468–72.
51. Wolber RA, Dupuis BA, Wick MR. Expression of c erb B-2 oncoprotein in mammary and extra-mammary Paget's disease. Am J Clin Pathol. 1991;96:243–7.
52. Beleznay KM, Levesque MA, Gill S. Response to 5-fluorouracil in metastatic extramammary Paget disease of the scrotum presenting as pancytopenia and back pain. Curr Oncol. 2009;5:81–3.
53. Takahagi S, Noda H, Kamegashira A. Metastatic extramammary Paget's disease treated with paclitaxel and trastuzumab combination chemotherapy. J Dermatol. 2009;8:457–61.
54. Cecchi R, Pavesi M, Bartoli L, Brunetti L, Rapicano V. Perineal extramammary Paget disease response to topical imiquimod. J Dtsch Dermatol Ges. 2010;8: 38–40.
55. Coldiron BM, Goldsmith BA, Robinson JK. Surgical treatment of extramammary Paget's disease. A report of six cases and a reexamination of Mohs micrographic surgery compared with conventional surgical excision. Cancer. 1991;4:933–8.
56. Hendi A, Brodland DG, Zitelli JA. Extramammary Paget's disease: surgical treatment with Mohs micrographic surgery. J Am Acad Dermatol. 2004;5: 767–73.

Malignant Conditions Including Squamous Cell Carcinoma and Rare Cancers

7

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Cancers of the anus, anal canal and anorectum are rare conditions comprising less than 2.5 % of gastrointestinal malignancies. However, the incidence has increased over the past several decades. In the United States there will be an estimated 5,820 new cases diagnosed in 2011, up from 4,660 new case in 2006 [1].

Anatomy

There are several important anatomic landmarks in the perianal region. The *anal verge* is the lowest portion of the anal canal or the external anal orifice. It overlies the intersphincteric groove and can be distinguished as the border between hair-bearing anal margin skin and non hair-bearing anoderm. The *anal margin* includes the skin 5 cm external, in a radial direction, to the anal verge that can be visualised by applying gentle traction to the buttocks. The *surgical anal canal* extends cephalad from the anal verge to the anorectal ring. The distal anal canal is lined by stratified squamous epithelium, while the proximal anal canal is lined by columnar epithelium similar to that of the rectum. In between is the *anal transition zone* demarcated distally by the *dentate line* and extends proximally 6–12 mm. This area is lined by basal, columnar and cuboidal cells, and the epithelium is cloacogenic, transitional or squamous.

Lymphatic drainage of the anal canal depends on the location in relation to the dentate line. The areas below the dentate line drain into the inguinal and femoral lymph nodes, whereas lesions above the dentate line are likely to drain to the perirectal and paravertebral lymph nodes, similar to rectal cancers.

Squamous Cell Carcinoma (SCC) of the Anal Margin

Anal margin is defined as the skin extending 5 cm external to the anal verge. Cancers in this area are less common that those within the anal canal. There are no large or prospective studies of anal margin tumours, and these lesions are often combined with anal canal cancers in larger series and trials. Anal margin tumours are uncommon. For example, they only comprise 7 % of anal tumours treated by the United Kingdom's Christie Hospital NHS Trust, reported in their large series of 254 anal cancer patients [2]. Wide local excision is the mainstay of treatment for small (≤2 cm), superficial well-differentiated tumours that can be removed with clear surgical margins, as continence can typically be maintained. With negative resection margins, outcomes are good.

Adjuncts such as chemotherapy and radiation have been used in more advanced disease

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or those that have recurred. Abdominoperineal resection should be reserved for those with locally recurrent disease after radiotherapy and/or chemotherapy. Patients with anal margin cancers that are not amenable to primary resection are thought to have less favourable outcomes [3]. However, in the few randomised trials on anal cancer that include anal margin tumours and report stratified results, anal margin cancers had similar outcomes to anal canal cancers [4, 5].

Several retrospective studies with small numbers of patients have been published looking at anal margin cancer specifically. Khanfir et al. report a 5-year locoregional control rate of 78 %, 5-year disease-free survival of 51 % and 5-year overall survival of 55 %. Only 35 % of these patients had chemotherapy [6]. Grabenbauer et al. describe patients with anal margin cancers, highlighting worse outcomes compared with anal canal cancers. These patients had a 50 % complete remission rate at 6 weeks after chemoradiation (compared with 92 % in anal canal tumours), 21 % local recurrence (compared with 8 % in anal canal tumours) and 5-year overall survival of 54 % (compared with 75 % for anal canal tumours). All patients in this series had chemoradiation (CRT) with 5-FU and mitomycin C [3]. Anal conservation rates are reported as 65 [7], 69 [3] and 80 % [6]. Chapet et al. report local control rates of 58 % in patients who had a local excision followed by irradiation and 64 % in patients with irradiation alone. 5-year overall survival in all 26 patients was 71 %. Half of these patients received chemotherapy [7].

Squamous Cell Carcinoma (SCC) of the Anal Canal

According to the United States' Surveillance Epidemiology and End Results (SEER) database, 50 % of patients with anal cancer have disease confined to the anus, 29 % have regional lymph node involvement or direct spread beyond the primary, and 12 % have metastatic disease, while 9 % have an unknown stage [8].

Risk Factors

Evidence indicates that the incidence of anal cancer is rising [9] possibly due to more prevalent human papillomavirus (HPV) infection, immunosuppression, anoreceptive intercourse and smoking [10]. In addition, history of other HPV-related gynaecologic neoplasms, particularly vulvar cancer, has been associated with increased incidence of anal SCC. Women with history of in situ of invasive gynaecological neoplasm have a 13-fold increase in anal cancer compared with expected rates [11].

Staging

Staging of anal cancer is done in accordance with the American Joint Committee on Cancer (AJCC); TMN staging system includes assessment of the size of the primary lesion, lymph node status and distant metastasis (Table 7.1). The primary tumour is evaluated by inspection and palpation to determine the size and location in relation to the anal verge and to determine if it involves the anal margin or anal canal. Presently, this staging classification does not take into account sphincter involvement, which may have functional and prognostic significance. Additional studies to further define the tumour anatomy and distribution are often necessary when the lesion is large or deeply invasive.

The extent of disease, including the presence or absence of metastatic lymph nodes, guides planning of radiation fields. Therefore, accuracy of pretreatment staging is important. Lymph node status is determined primarily by palpation of the groins with the addition of cross-sectional imaging to evaluate for deeper inguinal and pelvic nodes. There are no reliable size criteria for abnormal nodes. Enlarged groin nodes may be reactive and without malignant cells. Accuracy of clinical exam alone is disappointing since 44 % of lymph node metastases are less than 5 mm in diameter [12].

A chest x-ray or chest computed tomography (CT) and an abdominopelvic CT are often

Table 7.1 AJCC 7th edition staging system for anal cancer

T1	Tumour 2 cm or less		
T2	Tumour more than 2 cm but not more than 5 cm in greatest dimension		
T3	Tumour more than 5 cm in greatest dimension		
T4	Tumour of any size invades adjacent organ(s), e.g. vagina, urethra or bladder (invasion of the sphincter muscle(s) is not classified as T4)		
N0	No regional lymph node metastasis		
N1	Metastasis to perirectal lymph node(s)		
N2	Metastasis in unilateral internal iliac and/or inguinal lymph node(s)		
N3	Metastasis in perirectal and inguinal lymph nodes and/or bilateral internal iliac and/or inguinal lymph nodes		
M0	No distant metastasis		
M1	Distant metastasis		
Stage	**T**	**N**	**M**
I	T1	N0	M0
II	T2	N0	M0
	T3	N0	M0
IIIA	T1	N1	M0
	T2	N1	M0
	T3	N1	M0
	T4	N0	M0
IIIB	T4	N1	M0
	Any T	N2	M0
	Any T	N3	M0
IV	Any T	Any N	M1

included to evaluate for distant metastasis. Involved common iliac and periaortic lymph nodes are important to detect because while they are categorised as distant disease, it is possible to include them in the radiation fields.

Ultrasound

Endoanal ultrasound (EAUS) is a relatively inexpensive, safe and well-tolerated examination, which can be useful to evaluate the extent of local disease. There are limitations in patients with stenosis. In addition, the field of view is limited with an inability to assess distant mesorectal, inguinal or iliac nodes. In certain centres, EAUS is used to obtain two- or three-dimensional images to assess tumour location and spread within the anal canal and surrounding lymph nodes. The difficulty in assessing test characteristics such as sensitivity and specificity is that the majority of patients treated with chemoradiotherapy do not undergo pathological confirmation of node status. Some studies use a cut-off of 1 cm or larger for presumed positive lymph nodes [13]. Metastatic lymph nodes are typically round hypoechoic structures. Using size criteria alone, as mentioned above, may under-stage a significant percentage of patients.

MRI

Initial staging with magnetic resonance imaging may provide more useful information about local extent of pelvic disease, but it is more expensive and labour-intensive and may be contraindicated in certain patients (e.g. those with pacemakers). Anal canal tumours, when compared to the gluteus muscles, tend to have an intermediate signal intensity (SI) on T2-weighted images and isointense SI on T1-weighted images. Tumour involvement of the sphincter complex or urogenital structures can best be seen on T2-weighted MRI, as ill-defined intermediate SI infiltration or encasement [14]. Metastatic lymph nodes demonstrate similar signal intensity to the primary tumour. Perirectal lymph nodes with a maximum short-axis diameter of more than 5 mm and inguinal and pelvic sidewall nodes with a maximum short-axis diameter of more than 10 mm are often considered to contain carcinoma. Parikh et al. note similar limitations in both ultrasound and MRI with over-staging reactive lymph nodes and under-staging microscopic involvement. The European Society for Medical Oncology guidelines have recommended both EAUS and MRI in the primary staging of anal carcinoma. Ultrasound is felt to be more accurate for T stage, particularly early T stage, whereas MRI may identify more lymph nodes [13].

Several studies have evaluated the role of MRI in assessing the tumour before and after

CRT. It has been found to be useful in assessing the primary tumour size, signal intensity and infiltration of adjacent structures [15]. Tumour response was assessed by recording change in tumour size and signal intensity. After treatment, a decrease in tumour size accompanied by reduction and stability of the MR T2 signal characteristics at 1 year after CRT was associated with favourable outcomes. There are clear limitations to MRI including posttreatment oedema and scarring, but this tends to stabilise in T2-weighted SI and scar size after 1 year. Other studies have failed to identify predictive MRI features in the early posttreatment period for outcomes such as locoregional recurrence [16].

PET/CT

At diagnosis, ^{18}fluorodeoxyglucose uptake on positron emission tomography/computed tomography (FDG-PET/CT) is also used to evaluate lymph node status and distant metastases. FDG-PET/CT can be used for radiation therapy treatment planning by clearly defining sites of metabolically active tumour [17]. With FDG-PET/CT, the detection rate of non-excised tumours on initial examination was 93 % [18].

PET scan may provide additional information as a biomarker with higher maximum standardised uptake value (SUV) associated with an increased risk of nodal metastasis at diagnosis and worse disease-free survival. Patients with high anal tumour SUV (max) at diagnosis were at an increased risk of persistent or recurrent disease on post-therapy FDG-PET at 4 months. An SUV (max) $\geq$5.6 was associated with poorer disease-free survival [19]. In addition, PET/CT can upstage anal cancers and influence further management. In one study, 12.5 % of patients had a change in management based on PET/CT results, including 7.5 % ($n=3$) who were found to have FDG-avid inguinal lymph nodes that led to broader radiation fields. One patient had a FDG-avid periaortic node which was included in the radiation field, and one patient had a lung metastasis treated with metastasectomy [20].

Sentinel Lymph Node Biopsy

Sentinel lymph node biopsy (SLNB) has been studied as an adjunct to physical exam for evaluating for node-positive disease. 99mTc colloid is injected into the peri-tumoral tissue and lymphoscintigraphy performed. During the surgery, blue dye can also be used. The sentinel inguinal node is identified by a handheld gamma probe and dye visualisation. Multidirectional lymphatic drainage (mesenteric, iliac, inguinal) can occur in up to 56 % patients [21]. As many as 27 % of patients were found to have metastases in a lymph node that was not evident on clinical exam [22]. Other authors voice concerns over the use of sentinel lymph node biopsies to make decisions to omit inguinal radiation, particularly in patients with high-risk primary lesion. Several small series have reported 7–14 % rate of subsequent inguinal lymph node metastases within 2 years, despite negative SLNB on histopathology. Many of these patients did not have the inguinal lymph nodes included in the initial radiation fields. These were considered false-negative SLNB [21, 23]. De Jong's review of the literature included eight studies with a total of 143 patients. There was a 96.5 % detection rate of the sentinel lymph node, but the authors were unable to calculate a false-negative SLN detection rate. Further studies are needed before this can be broadly applied.

Prognostic Factors

The most important prognostic factors in anal cancer are thought to be T, N and M stages [24]. Locoregional control rates vary from 50 % at 3 years seen in the large (585 patients) UKCCCR randomised trial to 71.5 % at 5 years (88 % for stage I, 69 % for stage II, 77 % for stage IIIA and 60 % for stage IIIB) as reported in a group of 286 patients from France [25].

As the tumour size increases, there is a clear increase in local recurrence. Wright et al. in their series of 180 patients from Memorial Sloan-Kettering Cancer Center describe a 3-year locoregional failure rate of 15 % in T1/T2 patients compared with 42 % in T3/T4 patients

($p=0.0009$) [26]. Larger tumour size (higher T stage) is also associated with a decrease in survival (T1, 94 %; T2, 79 %; T3, 53 %; and T4, 19 %) [27].

The impact of nodal involvement on outcomes is weaker. The Radiation Therapy Oncology Group trial (RTOG 87-04) found those with node-positive disease had a higher colostomy rate (an indirect marker for local failure). Similarly, the European Organization for the Research and Treatment of Cancer (EORTC) trial comparing radiation alone to CRT showed significantly higher local failure rates ($p=0.0017$) and lower survival ($p=0.045$) in those with positive nodes, regardless of treatment arm, compared to node negative patients, but the extent or size of nodal spread did not influence prognosis [4]. In a 12-year review of 167 anal cancer patients, both increasing T stage (HR 1.7) and N stage (HR 1.47) were significantly associated with locoregional failure [28].

Certain pathological subtypes, such as basaloid subtype, and patients with human immunodeficiency virus have been shown to have a lower overall survival rate [28]. Additional factors found to be significant for worse prognosis on multivariate analysis included older age [3, 29] failure to complete radiation therapy [24], HIV-positive status [30] and dose intensity of chemotherapy less than or equal to 75 % [3]. Tobacco smoking has also been identified as a risk factor. Those who smoke tend to have the diagnosis of SCC at a younger age and have more frequent recurrence (32 % of smokers vs. 20 % of non-smokers) at a shorter interval. In addition, smokers had a significantly worse overall 5-year survival (45 % in smokers and 20 % in non-smokers; $p=0.05$) [31].

Surgical Therapy

Historically, surgical excision was the first-line treatment prior to the development of combined modality therapy championed by Nigro in the 1970s [32]. Before this sentinel publication, local excision was acceptable for lesions smaller than 2 cm with favourable pathological feature, confined to the mucosa or submucosa. The data on these small lesions is often skewed by the inclusion of those with anal margin cancer. Greenall et al. reported that 10 % of the anal canal lesions were amenable to local excision [33]. There was a 41 % rate of local recurrence and a 64 % overall 5-year survival. Those treated with abdominoperineal resection had a 38–71 % 5-year survival rate. With surgery alone, local pelvic or perineal recurrence accounted for 50–70 % of failures, and only 10–20 % died from distant metastases.

Current strategies favour combined modality therapy (CMT) first, with radical resection of anal squamous cell cancer reserved for those with persistent or recurrent disease after CMT, those who are unable to tolerate CMT and those who are not candidates for CMT.

Radiation Alone Versus Combined Modality Therapy

Radiation therapy results in an antitumour response in the majority of patients. Most protocols since Nigro's publications in 1974 have included chemotherapy with the radiation. However, external beam radiation therapy alone or in conjunction with brachytherapy was used in the 1980s and 1990s. With radiation alone, local control rates range from 61 to 100 % with overall 5-year survival rates of 50–94 % [34].

Two randomised controlled trials compare concomitant radiotherapy and chemotherapy to radiotherapy alone as definitive treatment for anal SCC.

The United Kingdom Coordinating Committee on Cancer Research (UKCCCR) randomised 585 patients with anal cancer to receive either 45 Gy in 20 or 25 fractions alone or with concurrent 5-fluorouracil during the first and last weeks of radiation and mitomycin on the first day of radiation [35]. They assessed for response at 6 weeks, and if there was >50 % tumour response, an additional boost of 25 Gy was given. Assessment of tumour response was done 2 months after completion of the boost. There was a higher incidence of early toxicity (within the first 2 months) with CMT, but the rates of late toxicity were similar. Early morbidities included leucopenia,

thrombocytopenia, skin reactions and gastrointestinal and genitourinary symptoms. There were no significant differences in perineal wound complications. There were similar rates of treatment-related morbidities necessitating surgery in both groups; these were included in the local failure analysis.

In both groups the majority of local failures occurred within the first 18 months. There was a significant reduction in the local failure rate in the CMT group (39 %) compared with the XRT-only group (61 %) at 3 years. The 3-year overall survival rate between the two arms was not statistically different (XRT only = 58 %, CMT = 65 %). The mortality rate from anal cancer at 3 years was significantly higher in the XRT group at 39 % versus 28 % in the CMT group ($p = 0.02$). Long-term follow-up at 12 years shows that these significant differences persist [36].

Similarly, the European Organization for the Research and Treatment of Cancer (EORTC) radiotherapy and gastrointestinal cooperative group demonstrated that the addition of chemotherapy to radiotherapy in patients with greater than 5 cm (≥T3) primary tumours or positive lymph nodes resulted in a significant increase in the complete remission rate from 54 % for radiotherapy alone to 80 % for CMT [4]. The locoregional control rate improved by 18 % at 5 years, and the colostomy-free rate increased by 32 % in those who received CMT. There was no significant difference in late side effects, although anal ulcers were more frequently observed in the combined-treatment arm. Despite better locoregional control and better progression-free survival in the CMT group, the survival rate remained similar in both treatment arms. Several significant prognostic factors were identified; nodal involvement, skin ulceration and male sex showed worse local control, and nodal involvement and skin ulceration showed worse overall survival. The size of the primary and percent circumference did not show any prognostic value nor did the location of the primary (canal vs. margin). Both UKCCCR and EORTC studies showed improved local control and decreased stoma rates with CMT compared to radiation alone without increased toxicity.

There have been several studies to evaluate the use of other cytotoxic chemotherapy agents in conjunction with radiation therapy. The most prominent has been cisplatin (CP). The RTOG 98-11 multicenter trial published in 2008 showed that the mitomycin C (MMC) group compared to the CP group had similar 5-year disease-free survival rates (60 % vs. 54 %; $p = 0.17$), overall survival rates (75 % vs. 70 %; $p = 0.10$), locoregional recurrence (25 % vs. 33 %) and distant metastasis (15 % vs. 19 %). The rate of colostomy was significantly lower for the MMC group (10 % vs. 19 % $p = 0.02$). There was, however, more haematological toxicity in the MMC arm [5].

Similarly, the Anal Cancer Trial (ACT) II failed to confirm any advantage of CP in the CRT regimen or 5-FU- and CP-based maintenance chemotherapy [37]. Other long-term studies show equivalent results in overall survival with MMC and CP. A study from Brazil showed that the overall colostomy rate was not significantly different with MMC versus CP. The 10-year overall survival and disease-free survival rates for the MMC group were 52 and 53 % and for the CP group 54 and 49 %, respectively ($p = 0.32$ and $p = 0.92$) [38].

Modifications of the radiation therapy include intensity-modulated radiation therapy (IMRT), which involves using PET/CT to defining high-, intermediate- and low-risk planning target volumes (PTV). Using treatment-planning software, the dose of radiation is tailored to provide graded doses of radiation to these different risk PTV areas. The high-risk PTV typically includes primary tumour and grossly positive nodal disease. Intermediate-risk PTV includes the internal iliac region inferior to the SI joint, the perirectal nodes as well as the high-risk PTV. The low-risk PTV area includes the inguinal nodes, external iliac nodes and the internal iliac nodes superior to the inferior edge of the SI joint. A recent small retrospective study has shown the benefits of IMRT with chemotherapy. With IMRT the duration of treatment is significantly shorter, requiring less frequent treatment breaks. In addition, when comparing IMRT to conventional RT, IMRT showed significantly better 3-year overall survival (88 % vs. 52 %), locoregional control (92 %

vs. 57 %) and progression-free survival (84 % vs. 57 %) [39]. The results of a prospective study, RTOG 0529, are awaited. A recent analysis of sites of locoregional failure from Memorial Sloan-Kettering Cancer Center concludes that inguinal and all pelvic nodal regions should be included in the PTV for IMRT, including the external iliac, internal iliac and presacral regions. The authors also recommend that common iliac nodes should be included in the radiation fields of patients with advanced T and N stage disease, based on 4 of 58 (7 %) common iliac node recurrences, three of which were not "in-field" in this subset of patients [26].

Management of Lymph Nodes

If the inguinal lymph nodes are found to be positive on physical exam or imaging and FNA confirms the finding, these nodes are then included in the radiation field. Many have advocated for routine prophylactic inguinal irradiation regardless of T stage, citing a 2 % 5-year inguinal lymph node recurrence rate for the prophylactic group compared to 16 % in those who did not get upfront inguinal radiation [40]. In patients who did not receive inguinal radiation, there was a 12 % rate of inguinal lymph node recurrence in those with T1 or T2 lesions and 30 % rate for T3 or T4 lesions. Many of the radiation treatment protocols used in the randomised trials include the inguinal nodes in all patients [5, 35, 38, 41].

Surveillance

The current guidelines for follow-up of patients with anal cancer after definitive CRT include serial digital rectal examination, with biopsy of suspicious lesions every 3 months beginning 8–12 weeks after completing CRT [42]. Cell death may continue up to 12 weeks after completion of CRT [43]. In addition, treatment-related ulcers may persist for 3–6 months [44]. Differentiation of treatment effect versus residual tumour can be challenging, and liberal use of biopsies is recommended. The Tru-Cut core biopsy needle can be used for sampling of deeper tissues in the ischiorectal fossa [3]. Most local recurrences, however, are apparent on physical exam, and a biopsy is obtained to confirm the diagnosis [35].

Follow-Up Imaging

The role of ultrasound in the follow-up of treated anal cancer is controversial. It is difficult to distinguish oedema and scar from persistent tumour on EAUS. In addition, this can be painful for patients with a relative anal stenosis. Some authors have recommended that waiting 16–20 weeks after radiation is sufficient to allow for resolution of oedema and improves the accuracy of the ultrasound imaging [45]. Serial exams can monitor for changes in the size of the scar, and this modality may add to routine clinical follow-up [46].

FDG-PET/CT, a non-invasive technique, has been studied to both determine residual disease and predict recurrence and survival. During post-treatment follow-up, FDG-PET/CT had, on a per examination basis, sensitivity for the detection of persistent or recurrent disease of 93 % and specificity of 81 % [18]. The 2-year cancer-specific survival was 94 % for those with a complete metabolic response (CMR) and 39 % for those with persistent FDG uptake post-CRT on PET/CT scan at a median of 2 months after completion of CRT, ($p = 0.0008$) [42]. CMR was associated with significantly improved progression-free and cause-specific survival compared with partial metabolic response. In fact, the results of the posttreatment FDG-PET/CT were more predictive of survival outcome than the pretreatment T stage. A similar predictive power of the PET response to radiation therapy has also been shown in a prospective study on 92 women with cervical cancer by the same researchers from Washington University in St. Louis, Missouri [47]. Another retrospective study of 48 patients with anal cancer showed a 5-year overall survival difference of 88 % in those with a CMR, 69 % for those with a partial metabolic response and 0 % in those with no metabolic response ($p < 0.0001$)

[48]. This study noted that 20 % of patients had coincident FDG-avid abnormalities that were not related to anal carcinoma—in three patients, separate primary malignancies were diagnosed. Studies on post-therapy PET/CT show that this modality is promising as a surveillance technique, but further confirmatory prospective data is needed to justify its routine use. Additionally, the timing of post-CRT PET/CT is thought to impact the specificity of this test, but at this point there is no data to clarify when PET/CT should be performed.

MRI has also been a part of follow-up but is reported in only a few studies. Stabilisation of the T2-weighted SI and scar size more than 1 year after CRT is associated with good outcomes in one small study of 15 patients [15]. With this modality it is important to establish a base line and look for stability of the images [14].

Outcomes

With varying CRT protocols using 5-FU/MMC and XRT, the complete response rates range from 72 to 95 %, with local failure of 25–39 % and overall 5-year survival of 58–84 % (Table 7.2). In multivariate analysis in the RTOG 98-11 trial, male sex ($p=0.02$), clinically positive nodes ($p<0.001$) and tumour size greater than 5 cm ($p=0.004$) were independent prognostic factors for worse survival. With the mitomycin-based treatment, local failure occurred in 13 %, regional failure rate was 7, and 25 % had distant metastasis at 5 years. Overall survival rates were less than 50 % at 4 years in those with tumour >5 cm and clinically positive lymph nodes [5].

Colostomy rate has been used as an end point for trials. Few studies distinguish the indication for the colostomy—whether it is created for tumour or treatment-related factors. Cumulative colostomy rates range from 4 to 23 % with standard CRT [5, 35, 37]. As many as 20 % of colostomies are created to deal with treatment-related effects [5, 35], and up to 10 % are created to deal with the presenting symptoms [37].

Complications

As mentioned above, radiation is associated with haematological toxicity in 60 % of patients that may interrupt treatment. Non-haematological toxicity rates may be reduced with the intensity-modulated radiotherapy technique [39]. Irradiation of the inguinofemoral region can lead to serious complications with acute and late toxicity. Acute toxicity includes epidermolysis with ulceration and superinfection of the skin, while late toxicity includes inguinal fibrosis, external genitalia oedema, neurogenic bladder, lower limb lymphedema, osteonecrosis of the femoral head, artery stenosis and soft tissue sarcomas. There was no difference in late toxicity rates observed

Table 7.2 Anal cancer outcomes after chemoradiation in randomised clinical trials

Study	*N*	Study comparison	Complete response (%)	Local/regional failure	Disease-free survival	Overall survival
UKCCCR [35]	283	XRT alone versus CMT	95	39 % at 3 years	–	65 % at 3 years
EORTC [4]	51	XRT alone versus CMT	80	33 % at 3 years*	–	58 % at 5 years*
ECOG/RTOG 87-04 [41]	146	XRT and 5-FU ± mitomycin C	92	–	73 % at 4 years	78 % at 4 years*
RTOG 98-11 [5]	324	CMT with mitomycin C versus cisplatin	–	25 % at 5 years	67 % at 3 years	84 % at 3 years
Brazil [38]	93	CMT with mitomycin C versus cisplatin	72	31 % at 5 years		61 % at 5 years

*Results from the mitomycin C groups displayed

between those receiving radiation alone and with the addition of 5-FU and MMC in both the EORTC and the UKCCR trials.

Quality of Life

Overall quality of life has been found to be good at a median of 51 months after CRT using the EORTC QLQ-CR29 and the global QLQ-C30 questionnaires in those who had a complete response [49]. Increased urinary frequency in 40 % of patients and some degree of faecal incontinence in 47 % of patients has been reported. More than half maintained an interest in having sexual relations, but 100 % of male patients had difficulty maintaining an erection. For women who maintained an interest in having sexual relations, 50 % reported having pain or discomfort during intercourse, and 100 % of men had difficulty maintaining an erection [49]. Another study that used EORTC questionnaires found that fatigue was the strongest predictor of impaired function-related quality of life [50].

Salvage Surgery

Salvage surgery is a curative approach to recurrent anal cancer after radiotherapy or chemoradiation. Almost always, an abdominoperineal resection (APR) is required, although scattered reports of local excision or low anterior resection can be found in the literature [51]. Between 17 % and 39 % of patients diagnosed with curable anal cancer will eventually require consideration of salvage APR for local control [2, 52, 53]. Not all patients with isolated local recurrence can be salvaged, as some will be unresectable when the recurrence is detected. In a large comprehensive series of anal cancer patients from a tertiary referral centre, Christie NHS hospital in Manchester, England, the surgical salvage rate for local recurrence was 82.7 % in those who had undergone chemoradiation and 71.4 % in those who had undergone radiation alone. Upon occasion, APR is performed because of the toxicity of radiation therapy [7]. In some cases recurrent or persistent disease cannot be ruled out, and these patients undergo APR [54].

Early detection of local recurrence can improve the chance for offering salvage surgery to the patient. Patients are examined carefully 3–6 months after chemoradiation or radiotherapy. Liberal use of examination under anaesthesia with biopsy is helpful as recurrences in the anal canal may be difficult to detect in patients who are often tender from treatment and whose examination findings can be confounded by radiation injury. Reported time to local recurrence after chemoradiation ranges from 12 to 21 months [55–58]. The recurrence is usually detected as a mass in 45–95 % of cases, and patients may complain of new anal pain or bleeding. The UKCCCR study found that recurrences were nearly always digitally palpable. In reported series of salvage APR for anal cancer, persistent disease comprises one-third to half of patients in nearly all series.

Preoperative planning is poorly described in published case series but is essential in planning an R0 resection. Renehan indicates that CT imaging, and more recently MRI, is used for preoperative planning. Preoperative planning is critical because many patients require a multivisceral resection (MVR) as part of salvage surgery. Most series report that 40–60 % of salvage patients require MVR [51–53, 56]. In one of the largest series ($n=95$) from France, Lefevre et al. report that 86 % of APRs for anal cancer included MVR, with the majority (70 %) comprising posterior colpectomy.

Salvage APR for anal cancer can be difficult and laborious due to bulky disease and fibrosis resulting from chemoradiation. The median blood loss ranges from 400 cc [57] to 1,000 cc [56], and operative duration has been reported as a mean of 4.6 [59] to 6 h [56]. In one series of 62 patients, three (8.5 %) patients developed profound bleeding during the resection that resulted in closing the abdomen with packs and reoperation the subsequent day [52].

Given the size of the soft tissue defect after APR and the difficulties with wound healing in an intensely radiated field, many authors describe myocutaneous flap closures of the perineal wound. The flap commonly used is the vertical rectus abdominis flap (VRAM). In many series,

nearly half of patients had a flap closure of the perineal wound [53, 56, 59]. Renehan et al. reported that nearly all cases are closed with a flap. Flaps do not completely prevent perineal wound complications, and in one series, all patients who had myocutaneous flap procedures developed perineal wound breakdown [58]. Lefevre et al. report no differences in perineal complications in the flap versus no flap groups, with a reoperation rate of 17 % in the VRAM group and 26 % in the group without VRAM. The benefit of VRAM was the decrease in time to perineal wound healing with a median time to healing of 19 days compared to 95 days in the group without a VRAM. There was a statistically significant difference in perineal hernia rates between groups, with no perineal hernias in patients with VRAM reconstruction.

Post-operative complication rates vary substantially. High rates (~70 %) are reported by Schiller et al. from Canada, Ferenschild et al. from the Netherlands and Lefevre et al. from France. Stewart et al. from the United States report an 18 % rate of late (>30 days) complications. These are predominantly perineal wound complications with rates of infection or dehiscence between 35 and 80 %. Renehan et al. describe that 66 % of perineal wound problems require over 3 months to heal, and Stewart et al. report a median time to perineal wound healing in all patients of 7 months.

Obtaining an R0 resection poses some challenges when performing salvage resection of recurrent or persistent anal cancer. Renehan et al. found that a positive resection margin was a risk factor for decreased overall survival in their report of 73 patients. R1 or R2 resections are reported in 8.5 [53] to 32 % [57] of cases, with many authors reporting rates close to 20 % [54, 56, 60]. Lefevre et al. argue that liberal use of the VRAM flap allows the surgeon to obtain wider margins on the tumour. The location of positive margins is not described in any of the literature.

Reported survival rates after salvage surgery range from around 30 [51, 60] to 64 % [53]. Survival rates are not improved with more recently published series suggesting that modern surgical practices are not improving outcomes. Predictors of survival are not consistent amongst reports, likely reflecting the small size of series—all include fewer than 100 patients and most include 40 or fewer patients (Table 7.3). Nilsson et al. found T3 or T4 tumours, persistent cancer, node-positive disease and older age associated with worse overall survival. Patients with persistent cancer had a 5-year overall survival of 33 % compared with 82 % in patients with recurrent cancer.

Table 7.3 Predictors of survival after salvage surgery for anal cancer

Study	Univariate analysis	Multivariable analysis
Akbari et al. [52]	Tumour size >5 cm	Persistent disease at salvage
	Adjacent organ involvement	Node-positive disease at salvage
Nilsson et al. [53]	Older age	None significant
	T3 or T4	
	Persistent disease at salvage	
	Node-positive disease	
Schiller et al. [57]	Charlson comorbidity	Charlson comorbidity
	Male sex	Male sex
	Lymphovascular invasion	Tumour size
	Histologic grade	
Stewart et al. [58]	Tumour size >5 cm	Node-positive disease
	Persistent disease at salvage	Positive margins
	Adjacent organ involvement	
	Node-positive disease	
	Positive margins	

Secondary failures after APR, unfortunately, are reported commonly. Reports range from 39 to 60 %. Most recurrences occur within 2 years [51], and many are locoregional diseases only. Eeson et al. also found that patients with HIV trended toward higher rates of recurrence (odds ratio 3.0; $p=0.08$) [60]. There is little information on the use of adjuvant therapy after salvage APR.

With high complication rates, modest survival benefits and high rates of recurrence after salvage APR, some authors question the utility

of salvage APR [60]; however, the procedure remains the only effective therapeutic option in these patients.

Metastasis

The role of hepatic resection for SCC remains poorly defined. In general, the development of distant metastasis portends a poor prognosis and there are no good therapeutic options. Salvage systemic chemotherapy has been used in SCC of the head and neck and is largely unhelpful with low response rates and short duration of response [61]. Despite being the most common site of distant metastasis in anal cancer, resection of hepatic SCC metastasis is uncommon [62]. Pawlik et al. published a multicenter study of 52 patients with SCC and liver metastasis that went on to liver-directed treatment [63]. In 27 of these, anal SCC was the primary. With a median follow-up of 18 months, nearly two-thirds developed recurrence. The median disease-free survival was 9.6 months. The liver was the most common site of tumour recurrence, with the majority also having extrahepatic disease as a component of failure. Those patients with hepatic metastases ≥5 cm in size and those with positive pathological resection margins tended to have increased risk of recurrence. Other factors, including presentation with synchronous distant disease, lack of response to chemotherapy, multifocal hepatic disease or bilateral liver disease, were not associated with risk of recurrence in this small study. Overall 5-year survival was 23 % after the hepatic metastasectomy. Overall survival was negatively impacted by synchronous disease, liver tumour size ≥5 cm and positive surgical resection margin. Longer disease-free interval may act as a marker of tumour biology. The authors of this study note that selection of appropriate patients for hepatic resection of metastatic SCC must be individualised and include an extensive evaluation of other sites of disease. Although the majority of patients recur, there is a subset of patients, as many as 25 %, that can achieve long-term survival. Since many recurrences occur systemically, improvements in survival will likely depend on the development of novel, more efficacious systemic chemotherapeutic agents.

Special Patient Subgroups: HIV, Transplant, and Systemic Lupus Erythematosus

HIV patients tend to be younger than non-HIV patients, male and present with early-stage disease. Most are on HAART therapy [30, 64]. Some studies have shown HIV+ patients tend to have lower complete response and overall survival rates [65], whereas others have shown the survival is not significantly worse in those with HIV [30]. Several studies agree that there are higher rates of treatment-related toxicity in those with HIV. These toxicities represent major clinical challenges and limit overall CRT dose and therefore can impact survival [65].

Solid organ transplant patients have an elevated risk of anal cancer compared to the general population (standardised incidence ratio 5.84; $p<0.001$), with an excess absolute risk of 9.6/100,000 person-years [66]. Patients with systemic lupus erythematosus are at increased risk for HPV-associated malignancies. A Danish cohort study noted a standardised incidence ratio (SIR) of 26.9 (95 % CI 8.7–83.4) with over 13 years of follow-up. There was also increased SIRs for vulvar, cervical and non-melanoma skin cancer [67].

Anal Adenocarcinoma

Anal canal adenocarcinomas are thought to arise from the ductal epithelium of anal glands at the level of the dentate line. These rare neoplasms are usually diagnosed after they have grown to a size that obliterates a definitive determination of the site of origin, and associated dysplasia in neighbouring epithelial cells is almost never described. It is estimated that adenocarcinomas comprise 16 % of anal canal neoplasms [68].

Information on the behaviour and possible treatment options can only be gleaned through several small case series. Beal et al. from

Memorial Sloan-Kettering Cancer Center in New York City report on 13 patients over a 12-year time period [69]. Basik et al. from Roswell Park Cancer Institute in Buffalo, New York, report ten patients over a 27-year time period [70]. Chang et al. report on 28 curatively treated patients over a 20-year time period from the MD Anderson Cancer Center in Houston, Texas [71]. Jensen et al. review 21 patients reported in a Danish national database over a 40-year time period [72]. Belkacemi et al. report on 82 patients collected over 25 years in a European Rare Cancer Network database [73]. In a less detailed analysis, the SEER database of the United States is used by Kounalakis et al. to describe the treatment and outcome of 165 patients over 16 years [74], and similarly Myerson et al. use the National Cancer Data Base (NCDB) of the American College of Surgeons to report on 213 patients over 1 year. While offering larger numbers, the disadvantage of the latter two databases is that distal rectal adenocarcinomas may not be reliably excluded from the analysis.

Treatment

The treatment approaches for anal adenocarcinoma are so variable that they are not consistent even within the same series. Radiation therapy is commonly used with or without surgery. Either local excision or abdominoperineal resection is chosen as the surgical approach, and criteria for choosing between these two options are not described. Some, but not all, reports include chemotherapy, and typically 5-FU-based chemotherapy is administered with scattered patients additionally receiving mitomycin C or less commonly cisplatin. Radiation therapy is usually administered via external beam in widely varying doses, up to 59 Gy. Seven (16 %) patients were treated with brachytherapy alone in the series by Belkacemi et al.

The overall impression of authors of recent series is that preoperative chemoradiation followed by radical resection of the anus is most likely to provide local control and possibly improved survival in patients with anal adenocarcinoma [69, 71, 75]. Chang et al. found radical resection to be the only predictor of overall survival in their multivariable analysis.

Prognosis

The most dismal survival data comes from Denmark where the 5-year survival was 4.8 %. Patients were older (median age 70 years) and most tumours were quite large (median 10 cm) at diagnosis. 62 % had distant metastases at presentation. Surgical treatment was utilised, but no mention of chemotherapy or radiation was made in this series. The best reported 5-year overall survival of 63 % was reported by Chang et al. amongst patients who underwent radical resection (APR). The associated median disease-free survival was 32 months. Nearly half (43 %) of curatively treated patients in this series underwent preoperative chemoradiation followed by APR. Patients who underwent local excision followed by external beam radiation had a 43 % 5-year overall survival and median disease-free survival of 13 months. Beal et al. report a 26-month median survival. Using SEER data from the United States, Kounalakis et al. found a 58 % 5-year survival in patients who underwent APR alone and 50 % 5-year survival in patients who underwent APR followed by XRT. Due to the nature of the database, the use of chemotherapy in these patients was unknown. Myerson et al. found a 5-year survival of 41 % in all patients. Basik et al. found a median survival of 29 months overall, and Belkacemi et al. found a 5-year overall survival of 39 %, which was improved amongst patients who had chemotherapy and radiation without surgery.

Recurrence

Local recurrence after various treatment approaches is common in most series. Local recurrence occurred in 35 % of patients over 5 years in the series from the Rare Cancer Network, a series in which only a minority of patients had an APR as part of treatment. The small series from Basik et al., Beal et al. and

Papagikos et al. found locally failure rates of 20, 38 and 54 %, respectively. Median time to recurrence was reported as 20 months in one series [75]. A few patients with local recurrences after local excision and radiotherapy underwent salvage APR with good results [69, 75].

Distant metastases are just as common as, if not more common than, local recurrence in most reported series. Chang et al. reported 43 % of patients developed distant metastasis. Fifty percent of patients in the series from Basik et al. recurred with distant metastases, and all of these recurrences were also associated with inguinal disease.

Prognostic Factors

As would be expected, earlier stage at diagnosis is associated with a better overall survival. In the report by Myerson et al., stages 0 through 2 had a 60.4 % 5-year overall survival, compared to stage 3 tumours with a 30 % 5-year survival. Belkacemi et al. found that T1 tumours were associated with a 72 % 5-year overall survival, compared to T2/T3 tumours, which had a 37 % 5-year overall survival. In their series, N2/N3 disease had a 13 % 5-year overall survival, compared to N0/N1 which had 46 % 5-year overall survival. Local recurrence was also statistically associated with higher T and N stage. Tumour size ≥4 cm was associated with worse 5-year overall survival in univariate, but not multivariable, analysis. Higher tumour grade was independently associated with overall (OR 3.65) and disease-free survival (RR 2.44). Tumour grade was also a predictor of disease-free survival in a multivariable analysis reported by Chang et al. Using SEER data, Kounalikis et al. found worse survival in older patients (OR 1.05), patients with node-positive disease (OR 3.77) and patients who had radiation alone compared to patients who had surgical treatment (OR 2.78).

Anal Melanoma

Anal melanoma is a rare tumour. The anorectum is the third most common site for a primary melanoma, and these melanomas represent 5 % or less of anal neoplasms [76, 77]. The incidence of anal melanoma is estimated at 1/1,000,000 for women and 0.7/1,000,000 for men [78]. In most series, women represent a higher proportion of patients than men. No increase in the incidence of anorectal melanoma has been detected over the past decades. Because of the rare nature of this tumour, information on its behaviour arises from small case series gathered over decades, which describe heterogeneous treatment approaches.

Most patients present with rectal bleeding and/or anal pain, and diagnostic delays are a common occurrence. In up to 50 % of patients reported in retrospective series, the lesion is initially diagnosed as a haemorrhoid [79, 80] and some are only diagnosed in pathological review of haemorrhoid specimens [81].

Anal melanomas vary in size at presentation. The majority of tumours are reported in the anal canal as opposed to the anal margin. Anorectal melanoma is thought to arise from melanocytic cells in the anal mucosa which can invade the lamina propria proximally into the rectal submucosa [82]. Many authors suspect that melanomas cannot arise from cells proximal to the transitional epithelium of the anal canal [78, 82]. A small proportion of melanomas grow in the rectum with some series specifying that intervening normal rectum is seen between the lesion and the dentate line. Occasionally authors describe patients with satellite tumour nodules in the distal rectum that has similarities to in-transit metastases in cutaneous melanoma [83, 84]. Histologic descriptions consistently show that 20–30 % of tumours are amelanotic [85–87]. Some tumours show ulceration and junctional activity. Spindled histology can be seen and explains why some tumours can be mistaken for sarcoma on histology [85, 86].

Anorectal melanoma can spread to the mesorectal, inguinal and pelvic lymph nodes. Spread to mesorectal lymph nodes is seen frequently, with a rate of 42–69 % in APR specimens [81, 83]. There is no AJCC staging system for anal melanoma. Authors typically classify patients as stage I for localised disease, stage II for locoregional disease (including inguinal and pelvic lymph nodes) and stage III for metastatic disease.

Details of diagnostic testing on presentation are not well described in the literature. A substantial proportion of patients present with metastatic disease from 20 to 60 % [83, 88, 89]. Case series show no obvious or consistent predictors of metastasis based on primary tumour characteristics. After diagnosis of anal melanoma, all patients should all have a metastatic work-up with cross-sectional imaging. Metastases are most commonly identified in the liver and by the lung. There are no reports on the utility of PET/CT for this disease. Tumour markers are not described except in the series by Ishizone et al. In this series of five case reports, serum 5-S-cysteinyldopa (5-S-CD) was reported in one patient; however, the results and utility of this test was not discussed.

Treatment

Surgery

Anal melanomas are curatively treated with either wide local excision or abdominoperineal resection (APR). Nearly all series show no survival differences between these two surgical approaches [90, 91] and therefore conclude that APR is appropriate only for tumours which cannot be merely excised due to sphincter invasion. This has been the conclusion of several authors of reviews on this disease [92, 93]. Even early-stage anal melanoma does not benefit from aggressive surgical resection. In a separate analysis of stage I patients from the nationwide registry of the Netherlands, patients who had an APR patients had the same rate survival as patients who had a local excision [94]. A few series show a survival advantage in the APR group. A small series of 19 patients from Korea showed an unusually prolonged median survival of 66 months in the 12 patients who had abdominoperineal resection and 11.2 months in the seven patients of the local excision group [87].

In terms of local control, APR is consistently found to be superior to a local excision. Many series report high local recurrence rates after wide local excision. Pessaux et al., in their series of 40 patients, found a 48 % LR rate after local excision compared to a 22.2 % local recurrence rate after APR. Ross et al. reported 32 patients from MD Anderson Cancer Center, finding local recurrence in 58 % of local excision patients compared with 29 % of APR patients; however, concomitant distant metastases were seen in 82 % of these patients [95]. Haitao reported a series of 57 cases from Beijing and found a 65 % rate of local recurrence in patients with a wide local excision statistically significantly higher than 15.6 % after APR. Belli found 45.8 % local recurrence rate after local excision versus no local recurrence after extended resection, but frequent distant metastases, seen in 69 % of patients after rectal resection. It must be emphasised that the impact of local recurrence on these groups of patients is outweighed by the frequent occurrence of metastatic disease. All patients with locoregional recurrence in the series reported from Antoniuk et al. showed metastatic disease within 6 months. Few case series comment on whether any patients die of anal melanoma without metastatic disease. Since case series are small, there are few tumour characteristics on physical examination, histology or imaging to allow the surgeon to determine whether a subset of patients could benefit from the decrease risk of local recurrence seen after an APR. Negative microscopic margins are advised whether the procedure is a local excision or an APR, but no studies have looked at this as a risk factor for recurrence. One study reported worse survival for R2 versus R0 resections, but no difference in 5-year survival between R1 and R0 resections [86].

Chemotherapy

Chemotherapy is used for treatment of patients with metastatic disease. Agents used typically include dacarbazine, nimustine, vinblastine and cisplatin. Response rates have not been favourable, except in case reports. The response rate to dacarbazine is reported to be 20 % at most. Adjuvant chemotherapy has also been reported [81, 87]. In 79 patients summarised over multiple case series from Japan, there was no significant difference in survival between 18 patients who received adjuvant chemotherapy and 15 patients who did not receive chemotherapy [85]. Biologic

therapy with interferon and interleukin-2 is also reported in limited series and with no clear success apart from isolated cases. One report from Memorial Sloan-Kettering documents complete regression of the primary and metastasis after cisplatin, temozolomide, doxorubicin and external beam radiotherapy, with 12 months of follow-up [96].

Radiation Therapy

Radiation therapy is occasionally reported in some patients from many series, typically used for local control of advanced tumours in patients with metastatic disease. One series from MD Anderson shows that local excision followed by 30 Gy of external beam radiation was adequate treatment and afforded a 31 % 5-year overall survival for patients with stage I and II disease [89]. Some authors report that a fraction of their patients underwent adjuvant radiotherapy, but selection criteria are not described [81].

Patterns of Recurrence

Most patients experience disease recurrence after curative procedures, and the majority of these (40–65 %) are distant metastases [81, 95]. Isolated local recurrence is infrequently reported and when present is seen in a small proportion of patients [81]. Inguinal node relapse is common, seen after treatment in up to 39 % of curatively treated patients [95]. In the report by Brady et al., 27 % of patients had an isolated inguinal recurrence [81].

Prognosis

The overall survival (OS) for patients with anal melanoma is poor. SEER reports a 5-year OS of 32 % for patients who present with local disease, 17 % for patients who present with regional disease and no survival in patients with metastatic disease. Median survivals are reported to be 12–18 months in most institutional series [79, 89, 95, 97] with some studies showing slightly more prolonged survivals of 22 months [85, 98]. Most reports comment upon a few patients who had long-term survival [76, 80, 82, 98].

Not surprisingly, advanced stage at presentation predicts worse prognosis [85, 90, 91, 94]. Brady et al. reported that amongst patients with resectable disease who underwent APR, those found to have uninvolved mesenteric nodes had significantly improved disease-free survival than those with positive mesenteric nodes (40 % vs. 11 %; $p<0.01$). For unclear reasons, patients have been found to have better survival in more recent series. Other markers that have been investigated show no consistent prognostic significance; however, it is difficult to draw firm conclusions due to a limited sample size in case series. Tumour thickness was associated with survival in the series of 36 patients from Memorial Sloan-Kettering by Wanebo, with patients who have lesions <2 mm experiencing long-term survival and poor prognosis (mean survival of 8 months) in patients with lesions 3–5 mm thick. Tumour thickness was also found to be associated with worse prognosis by Ballo et al. Tumour size <2 cm was associated with better survival in the series reported from the Swedish National Cancer Registry [78]. Ballo et al. also found that tumour size matters, reporting a disease-free survival of 66 % in tumours ≤4 cm versus 19 % in larger tumours ($p=0.04$). However, Pessaux et al. in an institutional review of 40 cases found no association between overall survival and tumour size or thickness. Depth of invasion is not reported consistently as a risk factor. The presence of melanin in the tumour is not predictive of better prognosis. Histologic factors such as mitotic rate or ulceration have not been investigated for prognostic significance.

Leiomyoma, Leiomyosarcoma and Gastrointestinal Stromal Tumour (GIST) of the Anus

Stromal tumours of the anus are extraordinarily rare. The collective literature on these tumours predominantly describes leiomyomas and leiomyosarcoma. Investigators in collaboration with John E. Skandalakis and Stephen W. Gray have

compiled and updated the case reports of these tumours from the worldwide literature extending back as far as 1881 [99, 100]. However, this literature combines tumours of the rectum and the anus. Leiomyomas and leiomyosarcomas are reported with similar frequency. The distinction between the malignant and benign smooth muscle tumours at any site of the body is dependent on histologic evaluation. Both tumours have a spindle cell appearance, with leiomyosarcomas showing pleomorphic cells, hyperchromatic nuclei and increased mitotic activity. Leiomyosarcomas and GISTs spread beyond the local site of origin to distant metastatic sites or the abdominopelvic cavity. Regional lymph node spread is uncommon.

In the most recent inventory of case reports, which tracks reports up until 1996, 432 leiomyomas and 480 leiomyosarcomas of the anus and rectum were identified [101]. Amongst cases where tumour site of origin was described precisely, 8.1 % of leiomyomas and 6.4 % of leiomyosarcomas were in the anus. Only 19 anal leiomyosarcomas have been reported in the world literature [102, 103]. Outcomes described for anorectal smooth muscle tumours are therefore heavily skewed by patients with rectal tumours. Anal tumours have been described at the internal anal sphincter, the anorectal junction and the anal verge. Over half of anorectal cases were larger than 5 cm at presentation. The peak age at diagnosis for anorectal leiomyomas was 40–59 years and for leiomyosarcoma was 50–69 years [101].

Overall, 20 % of anorectal leiomyosarcomas show metastatic behaviour. The liver is the most common site of spread, but metastases to the lung, bone and adrenals have also been described. Local disease recurrence is also common, occurring in 87 % of patients in the most recent evaluation of case series [101]. Some of these recurrences occur after a long (>5 years) disease-free interval. Based on small numbers over accumulated case series, the estimated 5-year survival rate is 37.5 %.

Authors recommend a complete excision, which in many cases amounts to an abdominoperineal resection [102, 104, 105]. Typically tumours >5 cm are bulky enough to require an APR, but there are no retrospective comparisons to support this. Leiomyomas should be widely excised, and a 2 cm margin has been recommended [100]. Careful attention to the margins may be important in preventing recurrence. Some leiomyomas are reported to recur as leiomyosarcomas [100, 101].

Anal GISTs are less common than smooth muscle tumours. Accurate reporting of GIST in the literature, which hinges on immunohistochemical staining for the tyrosine kinase receptor, *kit*, is reliable only in the past two decades. It is therefore possible that prior reports of anorectal leiomyosarcomas may in reality include unrecognised GISTs [106]. The largest series of anorectal GISTs by Miettenen et al., which combines the Armed Forces Institute of Pathology and University of Helsinki databases, includes only three anal tumours, comprising 2 % of the reported patients [107]. Two other recent reports can be identified in the literature [108, 109]. It is reasonable to suppose that the behaviour of this tumour likely follows the risk stratification criteria using tumour size and mitotic activity [110], though there is no evidence as of yet that substantiates this assumption. There is no retrospective data that can tease out whether a local excision may be adequate compared to an APR, though, as seen with the discussion on anal melanoma and anal adenocarcinoma, common sense dictates that an APR would be indicated when a local excision with negative margins is not possible.

References

1. Surveillance, Epidemiology, and End Results Program. SEER Stat Fact Sheets: Anal Cancer. http://seer.cancer.gov/statfacts/html/anus.html. Accessed 4 Jan 2012.
2. Renehan AG, Saunders MP, Schofield PF, et al. Patterns of local disease failure and outcome after salvage surgery in patients with anal cancer. Br J Surg. 2005;92:605–14.
3. Grabenbauer GG, Kessler H, Matzel KE, et al. Tumor site predicts outcome after radiochemotherapy in squamous-cell carcinoma of the anal region. Dis Colon Rectum. 2005;48:1742–51.
4. Bartelink H, Roelofsen F, Eschwege F, et al. Concomitant radiotherapy and chemotherapy is superior to radiotherapy alone in the treatment of locally

advanced anal cancer: results of a phase III randomized trial of the European Organization for Research and Treatment of Cancer Radiotherapy and Gastrointestinal Cooperative Groups. J Clin Oncol. 1997;15:2040–9.
5. Ajani JA, Winter KA, Gunderson LL, et al. Fluorouracil, mitomycin, and radiotherapy vs fluorouracil, cisplatin, and radiotherapy for carcinoma of the anal canal: a randomized controlled trial. JAMA. 2008;299:1914–21.
6. Khanfir K, Ozsahin M, Bieri S, et al. Patterns of failure and outcome in patients with carcinoma of the anal margin. Ann Surg Oncol. 2008;15:1092–8.
7. Chapet O, Gerard J-P, Mornex F, et al. Prognostic factors of squamous cell carcinoma of the anal margin treated by radiotherapy: the Lyon experience. Int J Colorectal Dis. 2007;22:191–9.
8. Abbas A, Yang G, Fakih M. Management of anal canal cancer in 2010. Oncology. 2010;24:364–9.
9. Johnson LG, Madeleine MM, Newcomer LM, et al. Anal cancer incidence and survival: the surveillance, epidemiology, and end results experience, 1973–2000. Cancer. 2004;101:281–8.
10. Daling JR, Madeleine MM, Johnson LG, et al. Human papillomavirus, smoking, and sexual practices in the etiology of anal cancer. Cancer. 2004;101:270–80.
11. Saleem AM, Paulus JK, Shapter AP, et al. Risk of anal cancer in a cohort with human papillomavirus–related gynecologic neoplasm. Obstet Gynecol. 2011;117:643–9.
12. Wade DS. Metastases to the lymph nodes in epidermoid carcinoma of the anal canal studied by a clearing technique. Surg Gynecol Obstet. 1989;169:238–42.
13. Otto SD, Lee L, Buhr HJ, et al. Staging anal cancer: prospective comparison of transanal endoscopic ultrasound and magnetic resonance imaging. J Gastrointest Surg. 2009;13:1292–8.
14. Parikh J, Shaw A, Grant LA, et al. Anal carcinomas: the role of endoanal ultrasound and magnetic resonance imaging in staging, response evaluation and follow up. Eur Radiol. 2011;21:776–85.
15. Koh DM, Dzik-Jurasz A, O'Neill B, et al. Pelvic phased-array MR imaging of anal carcinoma before and after chemoradiation. Br J Radiol. 2008;81:91–8.
16. Goh V, Gollub FK, Liaw J, et al. Magnetic resonance imaging assessment of squamous cell carcinoma of the anal canal before and after chemoradiation: can MRI predict for eventual clinical outcome? Int J Radiat Oncol Biol Phys. 2010;78:715–21.
17. Grigsby PW. FDG-PET/CT: new horizons in anal cancer. Gastroenterol Clin Biol. 2009;33:456–8.
18. Vercellino L, Montravers F, de Parades V, et al. Impact of FDG PET/CT in the staging and the follow-up of anal carcinoma. Int J Colorectal Dis. 2011;26:201–10.
19. Kidd EA, Dehdashti F, Siegel BA, Grigsby PW. Anal cancer maximum F-^{18} fluorodeoxyglucose uptake on positron emission tomography is correlated with prognosis. Radiother Oncol. 2010;95:288–91.
20. Engledow AH, Skipworth JRA, Blackman G, et al. The role of 18fluoro-deoxy glucose combined position emission and computed tomography in the clinical management of anal squamous cell carcinoma. Colorectal Dis. 2011;13:532–7.
21. Gretschel S, Warnick P, Bembenek A, et al. Lymphatic mapping and sentinel lymph node biopsy in epidermoid carcinoma of the anal canal. Eur J Surg Oncol. 2008;34:890–4.
22. De Nardi P, Carvello M, Canevari C, et al. Sentinel node biopsy in squamous-cell carcinoma of the anal canal. Ann Surg Oncol. 2011;18:365–70.
23. de Jong JS, Beukema JC, van Dam GM, et al. Limited value of staging squamous cell carcinoma of the anal margin and canal using the sentinel lymph node procedure: a prospective study with long-term follow-up. Ann Surg Oncol. 2010;17:2656–62.
24. Roohipour R, Patil SM, Goodman KA, et al. Squamous-cell carcinoma of the anal canal: predictors of treatment outcome. Dis Colon Rectum. 2008;51:147–53.
25. Tournier-Rangeard L, Peiffert D, Lanfond C, et al. Long-term results and prognostic factors of squamous cell carcinoma of the anal canal treated by irradiation. Cancer Radiother. 2007;11:169–77.
26. Wright JL, Patil SM, Temple LKF, et al. Squamous cell carcinoma of the anal canal: patterns and predictors of failure and implications for intensity-modulated radiation treatment and planning. Int J Radiat Oncol Biol Phys. 2010;78:1064–72.
27. Peiffert D, Bey P, Pernot M, et al. Conservative treatment by irradiation of epidermoid cancers of the anal canal: prognostic factors of tumoral control and complications. Int J Radiat Oncol Biol Phys. 1997;37:313–24.
28. Das P, Bhatia S, Eng C, et al. Predictors and patterns of recurrence after definitive chemoradiation for anal cancer. Int J Radiat Oncol Biol Phys. 2007; 68:794–800.
29. Nilsson PJ, Svensson C, Goldman S, et al. Epidermoid anal cancer: a review of a population-based series of 308 consecutive patients treated according to prospective protocols. Int J Radiat Oncol Biol Phys. 2005;61:92–102.
30. Hogg ME, Popowich DA, Wang EC, et al. HIV and anal cancer outcomes: a single institution's experience. Dis Colon Rectum. 2009;52:891–7.
31. Ramamoorthy S, Luo L, Luo E, Carethers JM. Tobacco smoking and risk of recurrence for squamous cell cancer of the anus. Cancer Detect Prev. 2008;32:116–20.
32. Nigro ND, Vaitkevicius VK, Considine B. Combined therapy for cancer of the anal canal: a preliminary report. Dis Colon Rectum. 1974;17:354–6.
33. Greenall MJ, Quan SH, Decoss J. Epidermoid cancer of the anus. Br J Surg. 1985;72:S97–103.
34. Eng C, Abbruzzese J, Minsky BD. Chemotherapy and radiation of anal canal cancer: the first approach. Surg Oncol Clin N Am. 2004;13: 309–20.

35. UK Co-ordinating Committee on Cancer Research. Epidermoid anal cancer: results from the UKCCCR randomised trial of radiotherapy alone versus radiotherapy, 5-fluorouracil, and mitomycin. UKCCCR Anal Cancer Trial Working Party. Lancet. 1996;348:1049–54.
36. Northover J, Glynne-Jones R, Sebag-Montefiore D, et al. Chemoradiation for the treatment of epidermoid anal cancer: 13-year follow-up of the first randomised UKCCCR Anal Cancer Trial (ACT I). Br J Cancer. 2010;102:1123–8.
37. James R, Wan S, Glynne-Jones R, et al. A randomised trial of chemoradiation using mitomycin or cisplatin, with or without maintenance cisplatin/5FU in squamous cell carcinoma of the anus. J Clin Oncol. 2009;27:18S.
38. Olivatto LO, Cabral V, Rosa A, et al. Mitomycin-C- or cisplatin-based chemoradiotherapy for anal canal carcinoma: long-term results. Int J Radiat Oncol Biol Phys. 2011;79:490–5.
39. Bazan JG, Hara W, Hsu A, et al. Intensity-modulated radiation therapy versus conventional radiation therapy for squamous cell carcinoma of the anal canal. Cancer. 2011;117:3342–51.
40. Ortholan C, Resbeut M, Hannoun-Levi JM, et al. Anal canal cancer: Management of inguinal node and benefit of prophylactic inguinal irradiation CORS-03 study. Int J Radiat Oncol Biol Phys. 2011;82(5): 1988–95.
41. Flam M, John M, Pajak TF, et al. Role of mitomycin in combination with fluorouracil and radiotherapy, and of salvage chemoradiation in the definitive nonsurgical treatment of epidermoid carcinoma of the anal canal: results of a phase III randomized intergroup study. J Clin Oncol. 1996;14: 2527–39.
42. Schwarz JK, Siegel BA, Dehdashti F, et al. Tumor response and survival predicted by post-therapy FDG-PET/CT in anal cancer. Int J Radiat Oncol Biol Phys. 2008;71:180–6.
43. Cummings BJ, Keane TJ, O'Sullivan B, et al. Epidermoid anal cancer: treatment by radiation alone or by radiation and 5-fluorouracil with and without mitomycin C. Int J Radiat Oncol Biol Phys. 1991;21:1115–20.
44. Borzomati D, Valeri S, Ripetti V, et al. Persisting perianal ulcer after radiotherapy for anal cancer. Hepatogastroenterology. 2005;52:780–4.
45. Giovannini M, Bardou VJ, Barclay R, et al. Anal carcinoma: prognostic value of endorectal ultrasound (ERUS). Endoscopy. 2001;33:231–6.
46. Tarantino D, Bernstein MA. Endoanal ultrasound in the staging and management of squamous cell carcinoma of the anal canal. Dis Colon Rectum. 2001;45:16–22.
47. Schwarz JK, Siegel BA, Dehdashti F, Grigsby PW. Association of posttherapy positron emission tomography with tumor response and survival in cervical carcinoma. JAMA. 2007;298:2289–95.
48. Day FL, Link E, Ngan S, et al. FDG-PET metabolic response predicts outcomes in anal cancer managed with chemoradiotherapy. Br J Cancer. 2011;105:498–504.
49. Provencher S, Oehler C, Lavertu S, et al. Quality of life and tumor control after short split-course chemoradiation for anal canal carcinoma. Radiat Oncol. 2010;5:41.
50. Weizel G, Hagele V, Wenz F, Mai SK. Quality of life outcomes in patients with anal cancer after combined radiochemotherapy. Strahlenther Onkol. 2011; 187:175–82.
51. Akbari RP, Paty PB, Guillem JG, et al. Oncologic outcomes of salvage surgery for epidermoid carcinoma of the anus initially managed with combined modality therapy. Dis Colon Rectum. 2004;47:1136–44.
52. Nilsson PJ, Svensson C, Goldman S, Glimelius B. Salvage APR in anal epidermoid cancer. Br J Surg. 2002;89:1425–9.
53. Mullen JT, Rodriguez-Bigas MA, Chang GJ, et al. Results of surgical salvage after failed chemoradiation therapy for epidermoid carcinoma of the anal canal. Ann Surg Oncol. 2006;14:478–83.
54. Lefevre JH, Parc Y, Kerneis S, et al. Abdominoperineal resection for anal cancer. Ann Surg. 2009;250:707–10.
55. Ferenschild FTJ, Vermaas M, Hofer SO, Verhoef C. Salvage abdominoperineal resection and perineal wound healing in local recurrent or persistent anal cancer. World J Surg. 2005;29:1452–7.
56. Schiller D, Cummings B, Rai S, et al. Outcomes of salvage surgery for squamous cell carcinoma of the anal canal. Ann Surg Oncol. 2007;14:2780–9.
57. Stewart D, Yan Y, Kodner IJ, et al. Salvage surgery after failed chemoradiation for anal canal cancer: should the paradigm be changed for high-risk tumors? J Gastrointest Surg. 2007;11:1744–51.
58. Papaconstantinou HT, Bullard KM, Rothenberger DA, Madoff RD. Salvage abdominoperineal resection after failed Nigro protocol: modest success, major morbidity. Colorectal Dis. 2006;8:124–9.
59. Van der Wal BCH, Cleffken BI, Gulec B, et al. Results of salvage abdominoperineal resection for recurrent anal carcinoma follow combined chemoradiation therapy. J Gastrointest Surg. 2001;5:383–7.
60. Eeson G, Foo M, Harrow S, et al. Outcomes of salvage surgery for epidermoid carcinoma of the anus following failed combined modality treatment. Am J Surg. 2011;201:628–33.
61. Colevas AD. Chemotherapy options for patients with metastatic or recurrent squamous cell carcinoma of the head and neck. J Clin Oncol. 2006; 24:2644–52.
62. Tokar EJ, Bobilev D, Zalmanov S, et al. Combined multimodal approach to the treatment of metastatic anal carcinoma. Onkologie. 2006;29:30–2.
63. Pawlik TM, Gleisner AL, Bauer TW, et al. Liver-directed surgery for metastatic squamous cell carcinoma to the liver: results of a multi-center analysis. Ann Surg Oncol. 2007;14:2807–16.

64. Oehler-Janne C, Huguet F, Provencher S, et al. HIV-specific differences in outcome of SCC of the anal canal. J Clin Oncol. 2008;26:2550–7.
65. Edelman S, Johnstone PAS. Combined modality therapy for HIV-infected patients with squamous cell carcinoma of the anus: outcomes and toxicities. Int J Radiat Oncol Biol Phys. 2006;66:206–11.
66. Engels EA, Pfeiffer RM, Fraumeni JF, et al. Spectrum of cancer risk among US solid organ transplant recipients. JAMA. 2011;306:1891–901.
67. Dreyer L, Faurschou M, Mogensen M, Jacobsen S. High incidence of potentially virus-induced malignancies in SLE arthritis. Arthritis Rheum. 2011;63:3032–7.
68. Myerson RJ, Karnell LH, Menck HR. The national cancer data base report on carcinoma of the anus. Cancer. 1997;80:805–15.
69. Beal KP, Wong D, Guillem JG, et al. Primary adenocarcinoma of the anus treated with combined modality therapy. Dis Colon Rectum. 2003;46:1320–4.
70. Basik M, Rodriguez-Bigas MA, Penetrante R, Petrelli N. Prognosis and recurrence patterns of anal adenocarcinoma. Am J Surg. 1995;169:233–7.
71. Chang GJ, Gonzalez RJ, Skibber JM, et al. A twenty-year experience with adenocarcinoma of the anal canal. Dis Colon Rectum. 2009;52:1375–80.
72. Jensen SL, Shokouh-Amiri MH, Hagen K, et al. Adenocarcinoma of the anal ducts: a series of 21 cases. Dis Colon Rectum. 1988;31:268–72.
73. Belkacemi Y, Berger C, Poortmans P, et al. Management of primary anal canal adenocarcinoma: a large retrospective study from the Rare Cancer Network. Int J Radiat Oncol Biol Phys. 2003;56:1274–83.
74. Kounalakis N, Artinyan A, Smith D, et al. Abdominal perineal resection improves survival for nonmetastatic adenocarcinoma of the anal canal. Ann Surg Oncol. 2009;16:1310–5.
75. Papagikos M, Crane CH, Skibber J, et al. Chemoradiation for adenocarcinoma of the anus. Int J Radiat Oncol Biol Phys. 2003;55:669–78.
76. Goldman S, Glimelius B, Pahlman L. Anorectal malignant melanoma in Sweden. Dis Colon Rectum. 1990;33:874–7.
77. Chang AE, Karnell LH, Menck HR. The national cancer data base report on cutaneous and noncutaneous melanoma. Cancer. 1998;83:1664–78.
78. Ragnarsson-olding BK, Nilsson PJ, Olding LB, Nilsson BR. Primary ano-rectal malignant melanomas within a population-based national patient series in Sweden during 40 years. Acta Oncol. 2009;48:125–31.
79. Homsi J, Garrett C. Melanoma of the anal canal: a case series. Dis Colon Rectum. 2007;50:1004–10.
80. Antoniuk PM, Tjandra JJ, Webb BW, et al. Anorectal malignant melanoma has a poor prognosis. Int J Colorectal Dis. 1993;8:81–6.
81. Brady MS, Kavolius JP, Quan SH. Anorectal melanoma: a 64-year experience at Memorial Sloan-Kettering Cancer Center. Dis Colon Rectum. 1995;38:146–51.
82. Wanebo HJ, Woodruff JM, Farr GH, Quan SH. Anorectal melanoma. Cancer. 1981;47:1891–900.
83. Cooper PH, Mills SE, Allen MS. Malignant melanoma of the anus: report of 12 patients and analysis of 255 additional cases. Dis Colon Rectum. 1982;25:693–703.
84. Duarte P, Ramos R, Vicente C, Casteleiro-Alves C. Anal melanoma with satellite implantations on the lower rectum. Rev Esp Enferm Dig (Madrid). 2011;103:49–50.
85. Ishizone S, Koide N, Karasawa F, et al. Surgical treatment for anorectal malignant melanoma: report of five cases and review of 79 Japanese cases. Int J Colorectal Dis. 2008;23:1257–62.
86. Pessaux P, Pocard M, Elias D, et al. Surgical management of primary anorectal melanoma. Br J Surg. 2004;91:1183–7.
87. Choi BM, Kim HR, Yun HR, et al. Treatment outcomes of anorectal melanoma. J Korean Soc Coloproctol. 2011;27:27–30.
88. Iddings DM, Fleisig AJ, Chen SL, et al. Practice patterns and outcomes for anorectal melanoma in the USA, reviewing three decades of treatment: is more extensive surgical resection beneficial in all patients? Ann Surg Oncol. 2010;17:40–4.
89. Belli F, Gallino GF, Lo Vullo S, et al. Melanoma of the anorectal region: the experience of the National Cancer Institute of Milano. Eur J Surg Oncol. 2009;35:757–62.
90. Podnos YD, Tsai N-C, Smith D, Ellenhorn JDI. Factors affecting survival in patients with anal melanoma. Am Surg. 2006;72:917–20.
91. Hai-tao Z, Zhi-xiang Z, Hai-zeng Z, et al. Wide local excision could be considered as the initial treatment of primary anorectal malignant melanoma. Chin Med J. 2010;123:585–8.
92. Singer M, Mutch MG. Anal melanoma. Clin Colon Rectal Surg. 2006;19:78–87.
93. Meguerditchian A, Meterissian S, Dunn KB. Anorectal melanoma: diagnosis and treatment. Dis Colon Rectum. 2011;54:638–44.
94. Roumen RMH. Anorectal melanoma in The Netherlands: a report of 63 patients. Eur J Surg Oncol. 1996;22:598–601.
95. Ross M, Pezzi C, Pezzi T, et al. Patterns of failure in anorectal melanoma. A guide to surgical therapy. Arch Surg. 1990;125:313–6.
96. Yeh JJ, Weiser MR, Shia J, Hwu W. Response of stage IV anal mucosal melanoma to chemotherapy. Lancet Oncol. 2005;6:438–9.
97. Moozar KL, Wong CS, Couture J. Anorectal malignant melanoma: treatment with surgery or radiation therapy, or both. Can J Surg. 2003;46:345–9.
98. Quan SHQ. Anal cancers: squamous and melanoma. Cancer Suppl. 1992;70:1384–9.
99. Skandalakis JE, Gray SW, Shepard D, Bourne GH. Smooth muscle tumors of the alimentary tract: leiomyomas and leiomyosarcoma: a review of 2525 cases. Springfield: Thomas; 1962.

100. Tarasidis G, Brown BC, Skandalakis IJ, et al. Smooth muscle tumours of the rectum and anus: a collective review of world literature. J Med Assoc Ga. 1991;80:685–99.
101. Hatch KF, Blanchard DK, Hatch GF, et al. Tumors of the rectum and anal canal. World J Surg. 2000;24: 437–43.
102. Thalheimer L, Richmond B, Lohan J. Leiomyosarcoma of the anal canal: case report and review of the literature. Am Surg. 2008;74:76–8.
103. Kumar GK, Chandra SS, Krishnan R. Local excision inadequate in the treatment of anal canal leiomyosarcoma. Saudi J Gastroenterol. 2010;16:226–7.
104. Wolfson P, Oh C. Leiomyosarcoma of the anus: report of a case. Dis Colon Rectum. 1977;20:600–2.
105. Kusminsky RE, Bailey W. Leiomyomas of the rectum and anal canal: report of six cases and review of the literature. Dis Colon Rectum. 1977;20: 580–99.
106. Nilsson B, Bumming P, Meis-Kindblom JM, et al. Gastrointestinal stromal tumors: the incidence, prevalence, clinical course, and prognostication in the preimatinib mesylate era. Cancer. 2005;103:821–9.
107. Miettenen M, Furlong M, Sarlomo-Rikala M, et al. Gastrointestinal stromal tumors, intramural leiomyomas, and leiomyosarcomas in the rectum and anus. Am J Surg Path. 2001;25:1121–33.
108. Tan GY, Chong CK, Eu KW, Tan PH. Gastrointestinal stromal tumor of the anus. Tech Coloproctol. 2003;7:169–72.
109. Nigri GR, Dente M, Valabrega S, et al. Gastrointestinal stromal tumor of the anal canal: an unusual presentation. World J Surg Oncol. 2007;5:20.
110. Fletcher CD, Berman JJ, Corless C, et al. Diagnosis of gastrointestinal stromal tumors: a consensus approach. Hum Pathol. 2002;33:459–65.

Rectocele Pathophysiology and Presentation

8

Karen P. Nugent

Definition

A rectocele is defined as the herniation or bulging of the posterior vaginal wall. The posterior vaginal wall and the anterior rectal wall are in direct apposition, and this bulging may be thought of as the rectum pushing anteriorly into the vagina.

Rectoceles may present in isolation but may also often be associated with pelvic organ prolapse (POP). POP describes the herniation of one or more of the pelvic organs including the uterus, the apex of the vagina, the bladder or the rectum from its normal anatomical position into or occasionally beyond the vagina.

Prevalence

Pelvic organ prolapse is common. In the United States, more than 200,000 operations are performed annually for one or more of the different presentations of pelvic organ prolapse, and numbers of women presenting for surgery for prolapse are increasing. The majority of pelvic organ prolapse is mild; however, prolapse beyond the vaginal introitus may occur in up to 5 % of cases. The Women's Health Initiative published in 2002 [1] suggested that in women aged between 50 and 79 years of age, up to 41 % of patients will present with some degree of POP, 18.3 % with a rectocele. However, the majority of these women are symptom free, and therefore, these prolapses may not be clinically significant. Several larger studies have shown a lifetime risk of symptomatic POP of around 11 % [2, 3]. As well as presenting with symptoms of bulge in prolapse, patients with pelvic floor disorders may also present with coexisting urinary or anal incontinence, and these symptoms often affect their quality of life, more than the prolapse itself.

The prevalence of rectoceles (either with or without POP) depends on whether they are asymptomatic or symptomatic. In a study from the 1980s, radiological rectoceles were seen in 81 % of nulliparous asymptomatic women who were less than 35 years old [4]. Forty-eight percent of the female patients had rectoceles that were greater than 1 cm in size, and one had one >2 cm. Coexistent intussusception was present in 50 % of women. A Swedish study invited female patients between 20 and 59 years of age for a gynaecological health examination [5]. 14.4 % of the population had a rectocele.

In 1997 the costs of repairing rectoceles in the United States (with or without other prolapse procedures) ran to $75 million [6, 7]. It is estimated that with an ever-increasing aging population, the healthcare costs for managing symptomatic rectoceles (along with other POP) will be enormous. The lifetime risk for undergoing a single operation for pelvic floor dysfunction is estimated to be 11.1 %, and up to 29 % of patients required reoperation [2].

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Aetiology

Predisposing factors associated with pelvic floor disorder and pelvic organ prolapse are advancing age, childbirth, obesity [8] and the menopause.

In a study from the South Australia Health Omnibus Survey, multivariate logistic regression showed that compared with nulliparity pelvic floor dysfunction was significantly associated with increasing relevance with caesarean section, then with a spontaneous vaginal delivery and with most importance with at least one instrumental delivery. Other pelvic floor morbidity associations were age, body mass index, coughing, osteoporosis and arthritis, and these resulted in reduced quality of life scores [9].

A further large study from Oxford Family Planning Association showed that the risk of hospital admission with prolapse increased with age, parity and weight. There were significant trends to increased risks with smoking status and obesity; the cumulative risk of requiring surgery increased following hysterectomy to 5.5 times higher in women whose initial hysterectomy was for genital prolapse as opposed to other reasons [10].

Although obviously multifactorial in nature, vaginal birth is the principal risk factor for developing POP, with proplapse occurring in 44 % of parous women and only 5.8 % of nulliparous women [5].

It appears that avulsion injury to the levator ani during childbirth may be the most significant structural damage; this is often also associated with fascial damage to the rectovaginal fascia as well as pudendal neuropathy (Fig. 8.1).

As stated before nulliparous women may also develop prolapse, 6.5 % in the Women's Health Initiative study [1] and the agreed associations above; there is also a clear genetic component as seen in twin studies [11].

Age is a significant risk factor [5]; the prevalence of rectocele is increasing with age from 15 % in the 30–39-year-olds age group to over 20 % in the 40–49-year-old age group and approximately 30 % by the time patients reach the age of 50–59. As previously mentioned, this study also confirms that not only is there an increase in age but prolapse occurs in 44 % of parous women, whereas the corresponding prevalence of amongst non-parous women is 5.8 %. Being overweight confers a significant increase in risk for developing a rectocele by 38 % if BMI is between 25 and 30 and by 75 % when the BMI is >30 [1].

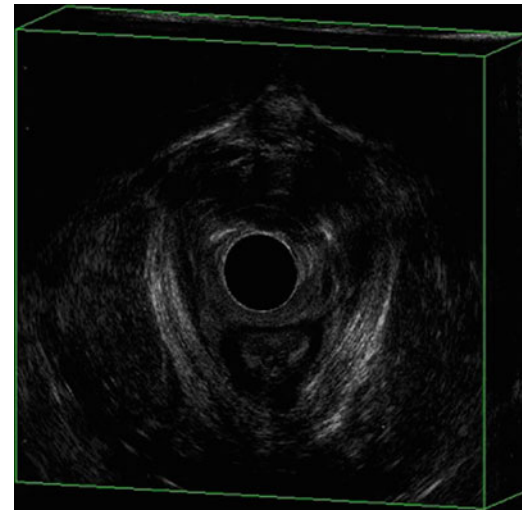

Fig. 8.1 Transvaginal scan showing symmetry and no damage to levator plate

Due to the multifactorial nature of pelvic organ prolapse and the fact that symptomatic rectoceles rarely occur in isolation, it is beholden upon a surgeon to understand which symptoms are most likely to be associated with the rectocele and therefore can be helped by correction of this anatomical abnormality. Patients who present with a rectocele may have a collection of both gynaecological and bowel symptoms as well as often having urinary symptoms too. The bowel symptoms may be constipation; obstructive defaecation (which includes an inability to empty); faecal incontinence, which is often passive or post-defaecatory leakage; pruritus ani; dyspareunia; and symptoms of bulge or pressure. In reality treatment of the rectocele in these patients with defaecatory problems may not only not improve their symptoms but make things worse. A very elegant paper by Pescatori [12] in 2006 described the many symptoms that a patient can suffer from when presenting with a primary problem of obstructive defaecation. He describes an iceberg syndrome, whereby the main presenting symptom

may underlie a multitude of many other problems. In his series of 100 consecutive constipated patients with a median age of 52, 54 of the patients had both mucosal prolapse and rectocele. However, 66 of the patients had at least 3 other problems including anxiety, depression, anismus and rectal hyposensation. Other associated problems included vaginal prolapse, neuropathy, enterocele and solitary rectal ulcer. In this series, the majority of patients were treated conservatively, and only 14 underwent any form of surgery. It is therefore important to take an excellent history to exclude or include other diagnoses and problems.

Dietz and Korda [13] in 2005 undertook a prospective study of 505 women presenting with symptoms to a tertiary urogynaecological clinic. They found 64 % of women had a rectocele. They examined the patients using transperineal ultrasound looking for herniation of the anterior anorectal muscularis and mucosa into the vagina. The symptoms strongly associated with posterior compartment descent were incomplete bowel emptying and digitation. Pain, chronic constipation and faecal incontinence were less strongly associated.

Pathophysiology and Anatomy of Rectocele

Although risk factors for POP can be identified, the specific events leading to a development of rectocele are poorly understood. It is likely that damage to a supportive layer of the fascia between the rectum and the vagina occurs. The origin of this fascia is in dispute, and a layer does not exist in isolation but as a component of other connective tissue that envelops the pelvic organs. Histologically the tissue is fibromuscular elastic layer which consists of dense collagen with course elastic fibres, smooth muscles and associated small blood vessels. Delancey's work [14] has divided this into three separate levels. From a craniocaudal direction, the upper third level or level one blends with the peritoneum of the culde-sac, the uterosacral ligaments and the base of the cardinal ligaments. The distal third of the vaginorectum, level 3, fascia blends with the perineum body. This has an important role in supporting the perineum with fascia attachments up to the ischiopubic rami and the urogenital diaphragm. For the middle third, level 2, the rectovaginal fascia extends laterally out to the fascia overlying the levator ani muscles.

The paper by Delancey in 1999 [14] was seminal in helping us understand the support system for the pelvic organs. Understanding these layers is essential to understanding what may go wrong. The support is multifaceted with connective tissue and striated muscles differing at different levels (Fig. 8.2).

The pelvic organ support system has an anterior compartment, middle compartment and posterior compartment. The vagina and uterus and endopelvic fascia lie within the middle compartment and attach themselves to the pelvic wall to separate the anterior and posterior compartments. Delancey undertook an extensive anatomical study of 42 fresh and 22 fixed cadavers to look at and define the posterior compartments structural anatomy relevant to rectoceles. The fixed specimens were used to study the overall arrangement of pelvic floor structures; however, the resistance of the posterior vaginal wall was studied in fresh cadavers not affected by fixation. As a result of this study, Delancey describes three different levels of rectum. The distal rectum lies in close apposition to the dense connective tissue of the perineal body at level 3. The perineal body (Fig. 8.3) represents the central connection between the two halves of the perineal membrane; when the distal rectum is subjected to force directed caudally, the fibres of the perineum membrane become tight and resist further displacement. This is necessary for evacuation so that there is substance or support for the waves of propulsion through the rectum to work against. Damage to the perineal body leaves the rectum more freely mobile and allows the distal rectum to prolapse downwards. The connections between the two halves of the perineal membrane extend cranially up from the perineal body for 2–3 cm whilst becoming progressively thinner towards the cranial margin. Above this level is level 2. The supporting structures of the middle portion

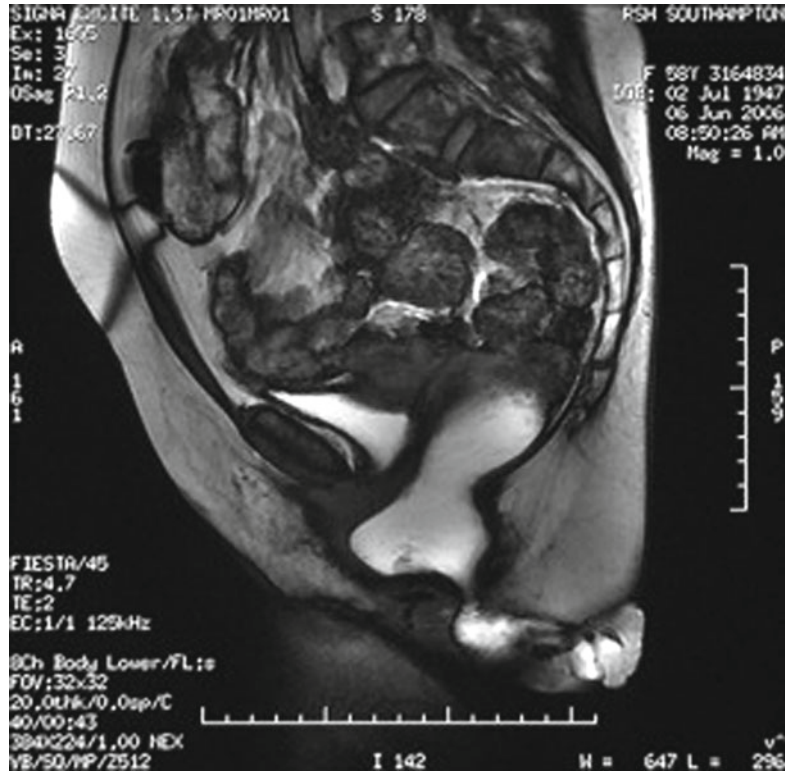

Fig. 8.2 Rectocele and its relation to pelvic organs

of the posterior vaginal wall (level 2) are attached on either side of the rectum to the inner surface of the pelvic diaphragm by a sheet of endopelvic fascia. These fascial sheets themselves attach to the posterior lateral vaginal wall producing a posterior vaginal sulcus on each side of the rectum. These endopelvic fascia sheets prevent the ventral movement of the posterior rectal wall.

The upper portion of the posterior vaginal wall (level 1) is attached to the pelvic wall by a sheetlike mesentery of the paracolpium. There are also coexisting muscular actions of the levator ani muscles which give additional support at level 2 and the upper surface of level 3. When these muscles contract, the connective tissue is elevated indicating that all these tissues work together to maintain support. The level 2 support is a balance between the fascia sheets and the muscles especially puborectalis; damage to either of these can result in a lack of vaginal wall support. The lower third is particularly supported by the dense connective tissue of the perineal body.

Damage to the different areas will produce different types of rectoceles. Defects in the mid vaginal support can give rise to a rectocele which may occur in the middle of the vagina despite normal muscle function and in the presence of an intact perineal body. Low or perineal rectoceles occur after damage to the perineal body and if the perineal body has been separated from the level 2 support above. It was Richardson who [15] began to suggest that identifying the particular defect is important in order to decide the type of repair required.

Results of rectocele repairs can only be examined when the type of damage or defect has been identified and specifically repaired. Identification of whether there is damage to the levator muscles or purely the fascia structures and also the preoperative findings of the state of the perineal body need to be taken into account. Indeed, in Richardson's paper he suggests that there are a variety of disruptions or tears within the layer called the rectovaginal septum. Probably the

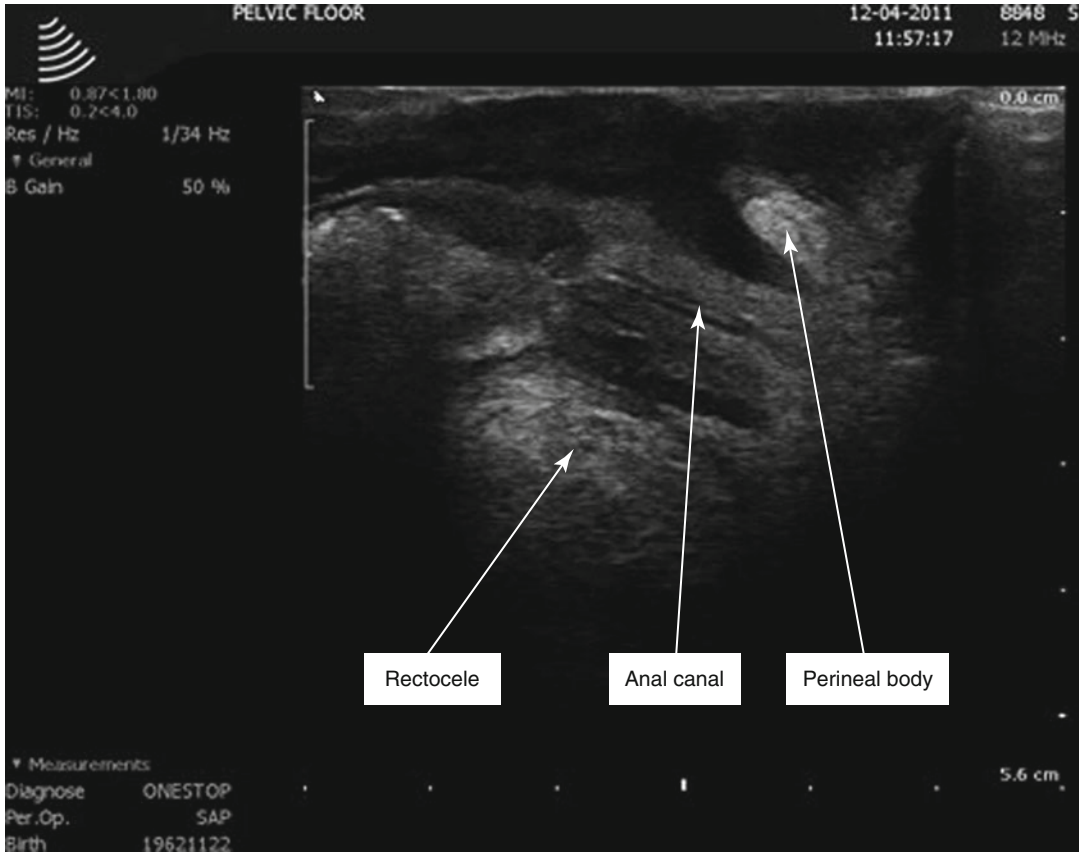

Fig. 8.3 Vaginal scan of perineal body and puborectalis

most common break accounting for rectoceles is a transverse separation immediately above its attachment to the perineal body; this tear usually yields a moderate-sized low rectocele that is a bulge that begins in an area just inside the introitus. A further equally common defect is a midline vertical defect that may indicate a poorly repaired episiotomy or obstetric injury. This tear may involve only the lower vagina but sometimes extends all the way up to the vaginal apex.

However, it is essential to remember that rectoceles are often present in nulliparous asymptomatic patients [4], and these patients may have no specific defect in their muscles or rectovaginal septum. This should remind us to question whether the presence of a rectocele is always correlated to the patients' symptomology and may help us understand why repair of a defect may not improve the patients' symptoms.

Clinical Presentation

As seen in the Shorvon paper [4], a small rectocele may cause no signs or symptoms. Patients may present with a bulge in the vagina which may or may not protrude through the introitus. The commonest colorectal symptoms in patients presenting with a rectocele are incomplete bowel emptying and digitation, which can be vaginal, perineal or rectal. They may also have a sensation of rectal fullness or pressure and a feeling of incomplete emptying after a bowel movement. For those with a rectocele through the introitus, the patients may complain of a dragging sensation. Pain, chronic constipation and faecal incontinence are also associated with significant fascial and pelvic floor defects [13].

Non-emptying is not a reliable symptom of a rectocele as seen by the Swedish study [5]; this

reported that 17.6 % of patients presenting with a rectocele had problems with emptying compared with 12 % in the non-rectocele group (P=NS). The symptom of non-emptying can be due to anismus or poor coordination of defaecatory effort – these patients may respond to biofeedback.

Other coexisting physical and functional problems make it difficult to be certain which symptoms are directly related to the rectocele [12]. Of more recent interest is the problem of occult rectal prolapse in patients with clinical rectoceles [16]. Investigations of symptoms may reveal that patients with a rectocele may have a rectorectal or rectoanal intussusception. This is defined as circumferential full thickness invagination of the rectal wall. Rectorectal intussusception is when the invagination remained within the rectum, rectoanal when it starts to come out of the anus. However, as described earlier, asymptomatic patients may have intussusception, and treating anatomical abnormalities may not help functional problems.

A series of 60 patients presenting with clinical rectoceles and defaecatory dysfunction had a selection of bowel symptoms [16]. They have one or more symptoms of constipation, incomplete evacuation with or without faecal incontinence, straining more than 25 % of the time and splinting. All 60 patients underwent defaecating proctography and of 20 (33 %) of them had significant intussusception and 10 % had anismus.

Patients with a rectocele may present with symptoms of a bulge, pressure or defaecatory problems. They should be properly evaluated as they may have many coexisting problems with their pelvic floor, and simple surgical treatment of their rectocele may not cure their symptoms.

References

1. Hendrix SL, Clark A, Nygaard I, Aragaki A, Barnabei V, McTiernan A. Pelvic organ prolapse in the women's health initiative: gravity and gravidity. Am J Obstet Gynecol. 2002;186(6):1160–6.
2. Olsen AL, Smith VJ, Bergstrom JO, Colling JC, Clark AL. Epidemiology of surgically managed pelvic organ prolapse and urinary incontinence. Obstet Gynecol. 1997;89(4):501–6.
3. Fialkow MF, Newton KM, Lentz GM, Weiss NS. Lifetime risk of surgical management for pelvic organ prolapse or urinary incontinence. Int Urogynecol J Pelvic Floor Dysfunct. 2008;19(3):437–40.
4. Shorvon PJ, McHugh S, Diamant NE, Somers S, Stevenson GW. Defecography in normal volunteers: results and implications. Gut. 1989;30(12):1737–49.
5. Samuelsson EC, Victor FT, Tibblin G, Svardsudd KF. Signs of genital prolapse in a Swedish population of women 20 to 59 years of age and possible related factors. Am J Obstet Gynecol. 1999;180(2 Pt 1):299–305.
6. Subak LL, Wing R, West DS, Franklin F, Vittinghoff E, Creasman JM, et al. Weight loss to treat urinary incontinence in overweight and obese women. N Engl J Med. 2009;360(5):481–90.
7. Subak LL, Waetjen LE, van den Eeden S, Thom DH, Vittinghoff E, Brown JS. Cost of pelvic organ prolapse surgery in the United States. Obstet Gynecol. 2001;98(4):646–51.
8. Kudish BI, Iglesia CB, Sokol RJ, Cochrane B, Richter HE, Larson J, et al. Effect of weight change on natural history of pelvic organ prolapse. Obstet Gynecol. 2009;113(1):81–8.
9. MacLennan AH, Taylor AW, Wilson DH, Wilson D. The prevalence of pelvic floor disorders and their relationship to gender, age, parity and mode of delivery. BJOG. 2000;107(12):1460–70.
10. Mant J, Painter R, Vessey M. Epidemiology of genital prolapse: observations from the Oxford Family Planning Association Study. Br J Obstet Gynaecol. 1997;104(5):579–85.
11. Altman D, Forsman M, Falconer C, Lichtenstein P. Genetic influence on stress urinary incontinence and pelvic organ prolapse. Eur Urol. 2008;54(4):918–22.
12. Pescatori M, Spyrou M, Pulvirenti d'Urso A. A prospective evaluation of occult disorders in obstructed defecation using the 'iceberg diagram'. Colorectal Dis. 2006;8(9):785–9.
13. Dietz HP, Korda A. Which bowel symptoms are most strongly associated with a true rectocele? Aust N Z J Obstet Gynaecol. 2005;45(6):505–8.
14. DeLancey JO. Structural anatomy of the posterior pelvic compartment as it relates to rectocele. Am J Obstet Gynecol. 1999;180(4):815–23.
15. Richardson AC. The rectovaginal septum revisited: its relationship to rectocele and its importance in rectocele repair. Clin Obstet Gynecol. 1993;36(4):976–83.
16. Thompson JR, Chen AH, Pettit PD, Bridges MD. Incidence of occult rectal prolapse in patients with clinical rectoceles and defecatory dysfunction. Am J Obstet Gynecol. 2002;187(6):1494–9; discussion 9–500.

Rectocele Treatment

9

Bruno Roche

Rectoceles are defined by a herniation of the rectal wall through a defect in the posterior rectovaginal septum in direction of the vagina. They are prevalent in women, but some rare cases of herniation of the rectum through the Denonvilliers' fascia have also been reported and treated in men [1]. This anatomical abnormality may occur for different reasons:

- The weakness of the rectovaginal septum may be congenital as in young nulliparous women.
- The weakness may be acquired postobstetrically in normal and traumatic deliveries. During deliveries the levator ani and puborectalis sling are dramatically stretched and in some instances disrupted (Fig. 9.1). The introitus then has a different appearance comparing nulliparous and primiparous women (Fig. 9.2).
- A rectocele may also be the end result of chronic straining because of constipation.

Small rectoceles can be found in nulliparous women [2] and most of the time are asymptomatic.

Rectoceles can be classified applying different criteria, for example, according to their position—low, middle or high [3]—and/or their size: small (<2 cm), medium (2–4 cm) or large (>4 cm) [2]. Size is measured anteriorly from a line drawn upward from the anterior wall of the anal canal on proctography. The most useful classification organizes the rectocele and its treatment into three clinical stages at straining during defaecating proctography [4, 5] (Table 9.1).

Furthermore, the length of the anal canal seems to play an important role in the development of incontinence and urgency.

If the anal canal on lateral view is symmetric and if the anterior and posterior sides are approximately equal in length, a rectocele may result from chronic constipation or dyschezia. If the anterior part is short resulting in an anal asymmetry, the patient fills the rectocele with stools, and despite a good anterior displacement of the puborectalis sling, it is unable to retain faeces. This results in incomplete exoneration, persistent need to pass stools or even marked incontinence.

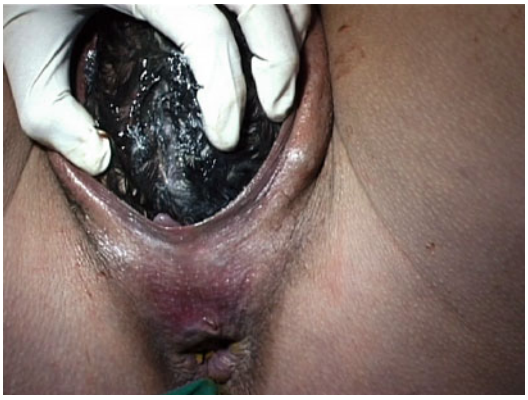

Fig. 9.1 Expulsion time: the levator ani and puborectalis sling are stretched

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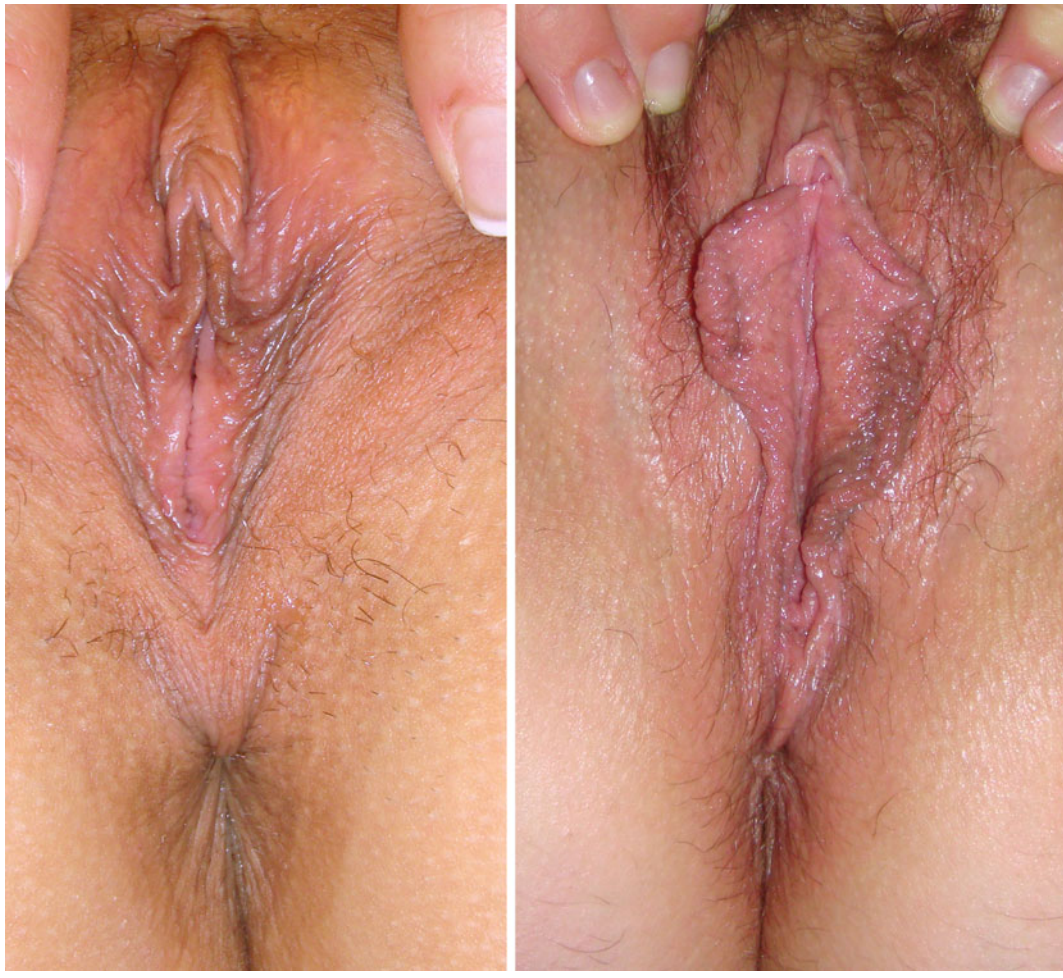

Fig. 9.2 Introitus aspect in nulliparous and multiparous

Table 9.1 Classification of rectoceles

Type I	Digitiform rectocele or single hernia through the rectovaginal septum
Type II	Lax rectovaginal septum large sacculation Anterior rectal mucosal prolapse Deep Douglas pouch Frequently associated with an enterocele
Type III	Rectocele associated with invagination and/or prolapse of the rectum frequently associated with an enterocele

Small rectoceles rarely produce symptoms [6, 7]. Large rectoceles may cause obstructed defaecation, constipation, pain and bleeding due to ulceration [8]. Sometimes they interfere with sexual function. Perineal pressure or vaginal digitation is often described as a measure taken by the patient to help defaecation. Incomplete evacuation of the rectoceles on defaecating proctography is an important finding [5]. In case of major postpartum damage of the perineum, we can observe sphincter lesions, perineal body damage and stretching and/or rupture of the puborectalis. These ruptures can occur at the insertion point of the muscle. Clinically, the patient presents with a deviation of the anus on the healthy side and a large introitus with a wide vagina. One can often demonstrate evidence of puborectalis damage with clinical exam and ultrasound (Fig. 9.3).

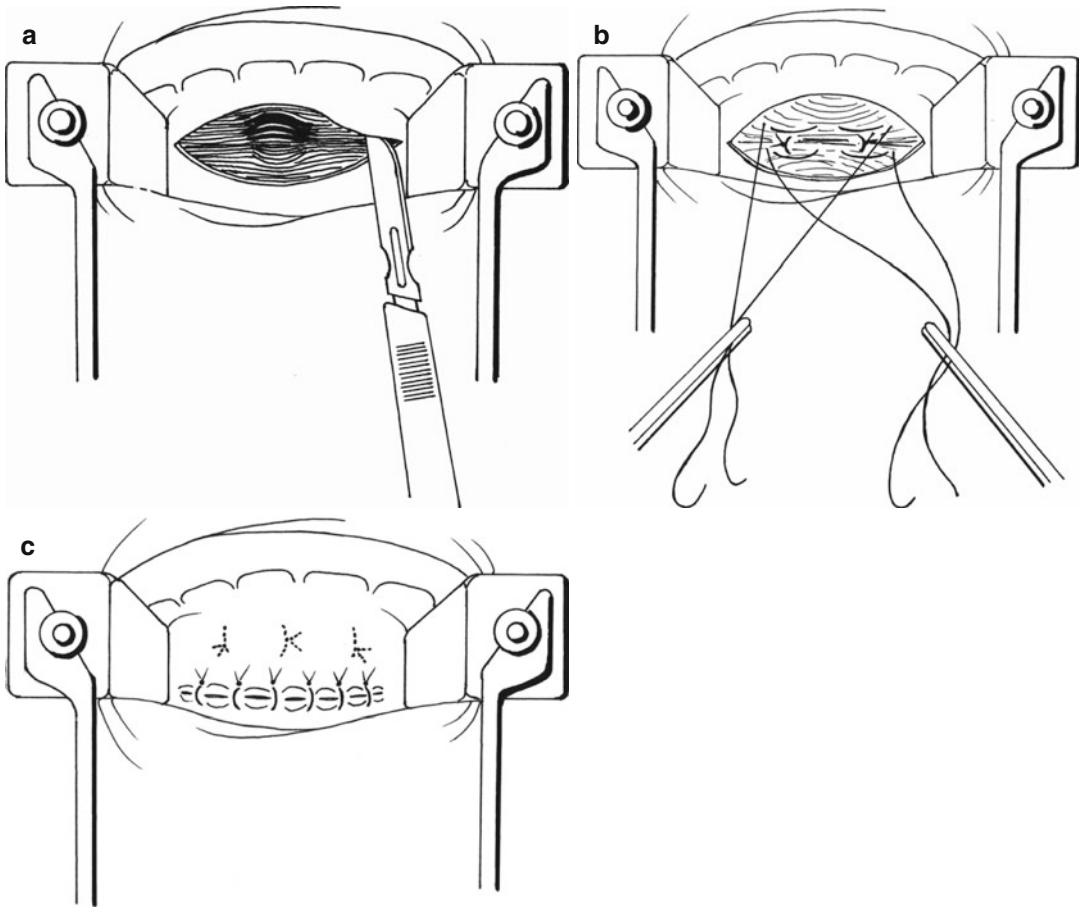

Fig. 9.3 Type I rectocele treatment according Sullivan-Sarles procedure: (**a**) Incision of anterior rectal mucosa above the dentate line. The rectal wall is exposed to the apex of the rectocele. (**b**) The rectal muscle is plicated, using a series of vertically placed sutures to reinforce the rectal wall. (**c**) Redundant mucosa is excised and the defect closed

Treatment

Medical

Once the diagnosis is confirmed, a high-fibre diet (25–35 g/day) plus ingestion of 2–3 l of non-caffeinated, nonalcoholic fluids per day is recommended as initial conservative treatment. Other conservative methods of treatment such as pelvic floor exercises, electrical stimulation of the pelvic floor muscles and the use of supportive devices such as pessaries within the vagina are of limited use [9]. If control of the patient's symptoms by conservative treatment is suboptimal, the authors will consider surgical therapy.

Surgical

Surgical repair should only be carried out on carefully selected symptomatic patients or in the context of any coexisting pelvic floor abnormality. Surgical indications for a symptomatic rectocele repair include the presence of obstructive defaecation symptoms, lower pelvic pressure and heaviness, prolapse of the posterior vaginal wall, pelvic relaxation or enlarged vaginal hiatus. The best results occur in patients who need to vaginally digitate [10] rather than in those needing to apply pressure to the perineum or digitate rectally [11]. Barium trapping on defaecography is also often used as a selection criterion [12]

Table 9.2 Surgical policy in the treatment of rectoceles

Type I		Endorectal repair (Sullivan-Sarles, STARR)
	+Incontinence	Sullivan-Sarles + transverse plication of internal sphincter
Type II		Perineal or vaginal repair
	+Incontinence	Sphincter repair
Type III		Double abdomino-vaginal approach according to Zacharin.

although it is not felt to be directly associated with defaecatory dysfunction [5]. An abnormal transit study with coexisting constipation might predict a less favourable outcome post rectocele repair [12].

Several approaches have been described to repair rectoceles:

In order to tailor the surgical procedure in accordance with the lesions observed and their functional complications, we have prospectively applied the following policy (Table 9.2):

1. The endorectal approach with stitches or stapler [10, 13–23]
2. The transvaginal or transperineal approach [12, 24–27]
3. The abdominal or abdominoperineal approach [28–30]

The surgical treatment should be tailored to correct the type of rectocele, the associated symptoms and any other concomitant pelvic floor disorders.

Type I Digitiform Rectocele or Single Hernia Through the Rectovaginal Septum

Type 1 rectoceles are treated by the endorectal approach, which was described by Sullivan et al. [16] and Sarles et al. [14, 31] and is usually carried out with the patient in the prone jackknife position. We prefer a classic gynaecologic position with the advantage of less anaesthetic risk and a shorter operating room time while the patient position does not need to be changed. The anterior rectal submucosa above the dentate line is infiltrated with a solution of local anaesthetic and a vasoconstrictor. A flap of mucosa is raised off the rectal circular muscle to expose the rectal wall as far as the apex of the rectocele. The rectal muscle is then plicated, using a series of five to eight vertically placed sutures to reinforce the rectal wall. Redundant mucosa is excised and the defect closed [14, 16]. Some surgeons prefer to plicate the muscle with horizontal mattress sutures [10, 32] (Fig. 9.3a–c).

In the presence of an intact but weak sphincter and a short anterior anal canal, the longitudinal plication can be completed by two or three double-V transverse sutures to lengthen the anal canal. This reinforces the occlusion mechanism and improves incontinence.

An anterior Delorme's procedure has been described as another anal approach to rectoceles, especially with an associated mucosal prolapse [33] or rectal invagination [34].

Endorectal procedures done with mechanical sutures were already described in 1993 by different authors [21, 22].

This likely inspired Longo and partners in the description of the STARR procedure. The stapled transanal rectal resection (STARR) procedure has been proposed for large rectoceles with intrarectal intussusception and a history of obstructed defaecation syndrome. Intrarectal intussusception described on defaecating proctography may suggest a mechanical obstruction to defaecation. Unfortunately, this has never been clearly proven as a direct cause of obstructed defaecation because the proctograms used in demonstrating this theory have always shown an empty rectum; thus intussusception cannot be advocated as the cause of obstruction.

The objectives of the STARR procedure in these patients are:

1. Removal of the prolapsing rectum and restoration of the normal anatomy
2. Re-establishing the continuity of the rectal muscular wall in order to regain normal rectal capacity and compliance
3. Anatomical correction of the rectocele or posterior colpocele

The largest prospective multicentre trial containing 90 patients undergoing the STARR procedure

for treatment of outlet obstruction caused by the combination of intussusception and rectocele has shown encouraging early results [35–37]. All patients had significant improvement in constipation symptoms without affecting continence, and postoperative defaecating proctography showed the disappearance of both the intussusception and rectocele [17].

Severe complications, however, have been reported, including bleeding, faecal urgency, incontinence, stenosis, dramatic chronic pain, constipation [38] and rectovaginal and enteral fistulas [39, 40].

In our experience these complications are very difficult to treat. They result in an important negative influence on patient quality of life following treatment of a benign disease. The presence of an entero- or sigmoidocele at rest is a contraindication to the use of this technique.

Type II Large Sacculation of Lax Rectovaginal Septum

Type II rectoceles may be treated by transvaginal or transperineal approaches, with or without the use of prosthetic materials to support the repair.

If a transverse perineal incision is used, the whole of the rectovaginal septum is exposed, separating each structure to demonstrate the levator ani, which is plicated using a series of horizontal mattress sutures. The interposition of mesh at this site has been reported [41]. Major complications such as erosion and infections may occur between 5 and 25 % [42–46]. It has been proposed that this approach is limited as it does not allow enough access to the upper part of the rectovaginal septum and to the pouch of Douglas. However, this approach can be useful in cases with a concomitant sphincter repair.

The transvaginal approach with an extensive posterior colpomyorrhaphy is a technique that has traditionally been preferred by gynaecologists and performed in the lithotomy position. A Lone Star retractor is useful, and following local anaesthetic and vasoconstrictor infiltration, the vaginal mucosa is incised transversely at the posterior hymen level and mobilized as far as the cervix or vaginal vault. The peritoneum at this point is mobilized and pushed up, and a Douglasorrhaphy may be performed. The levator ani and puborectalis muscles are dissected out and approximated at the midline with sutures. The first three sutures include the rectal muscle superficially 1–1.5 cm below the level of the first stitch into the puborectalis muscle. The rectal wall is then lifted and an anterior rectal mucosal prolapse is prevented (Fig. 9.4a–e). When the vaginal vault is prolapsing, a sacrospinal fixation may also be added [3, 47, 48]. In these cases, a colpopexy to the sacrospinal ligament may also be carried out.

If there is sphincter rupture or stretching, a sphincteroplasty may also be performed. One should note that an endovaginal approach is necessary in order to repair cases involving a ruptured puborectalis muscle (Fig. 9.5).

In our centre, we treated 32 young women (35.5 years), all presenting with a rectocele and associated anal incontinence. The main complaints were anal incontinence in all patients with the average Wexner score of 12.4, sexual problems in 23 patients, dyschezia in 11 and colophony in 8. The puborectalis was repaired in 20 cases on the right side, 9 on the left and 3 bilaterally (Fig. 9.6). Clinically, the rectocele was corrected in all cases with improvement in the Wexner incontinence score which dropped from 12.4 to 2.7. As well, patients showed improvements in sexual problems (10/23), dyschezia (4/11) and colophony (6/8). In this sense we can agree with Beck's [49] conclusion stating that: "Overall, surgical correction success rates are quite high when using a vaginal approach for rectocele correction. Vaginal dissection results in better visualization and access to the endopelvic fascia and levator musculature. This allows for a more precise and solid anatomical correction."

Direct comparison of the transanal and transvaginal approaches in a prospective study of 30 female patients with symptomatic rectoceles, excluding patients with compromised anal sphincter function or other symptomatic prolapse, was carried out by Nieminen et al. [27]. There were 15 patients in each study group. At 1 year follow-up, 73 % (11/15) showed improvement

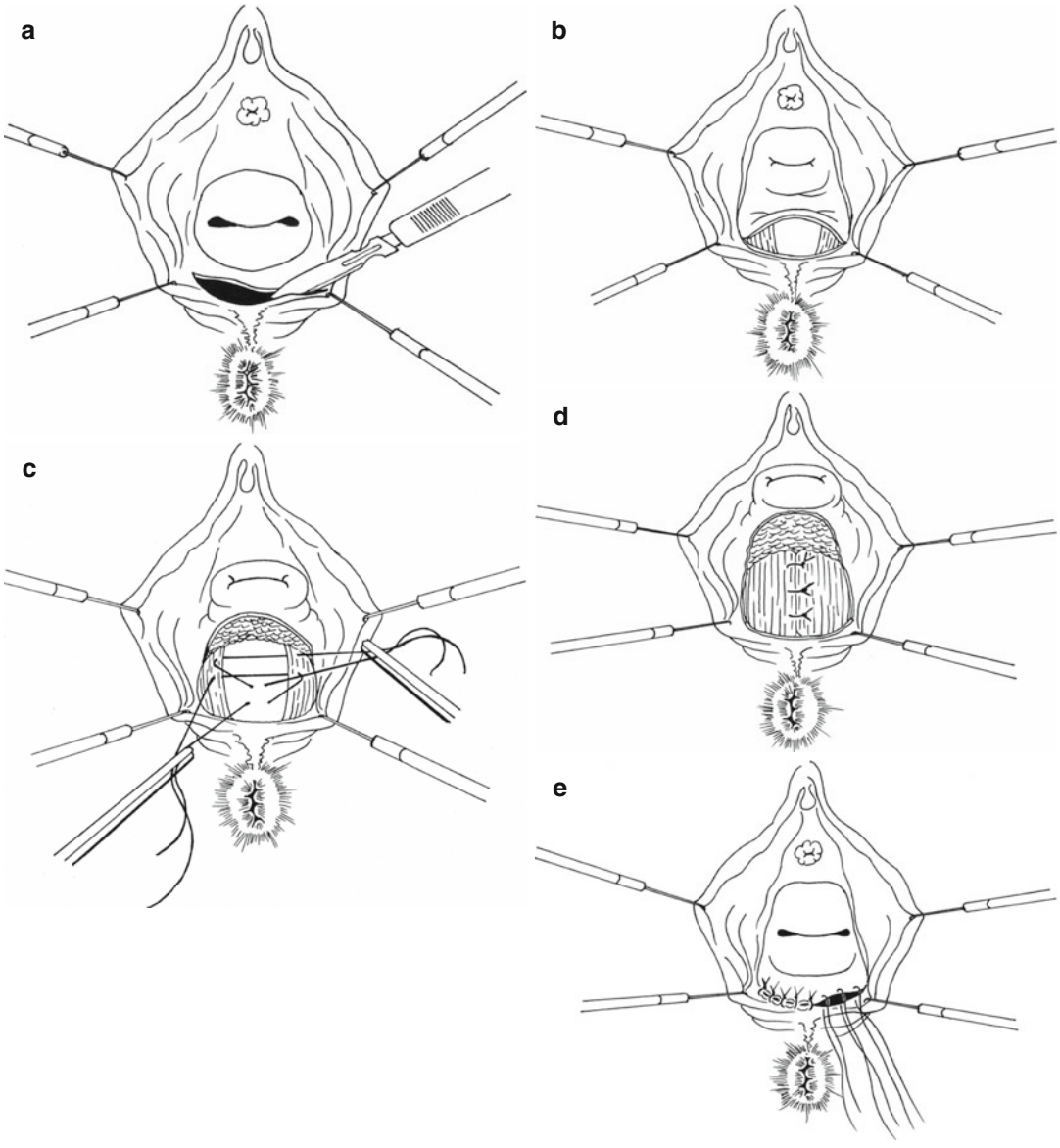

Fig. 9.4 Type II rectocele treatment. (**a**) Transversal incision of vaginal mucosa. (**b**) Dissection of levator ani and puborectalis muscles. (**c** and **d**): Levator ani suture with rectal wall pexy at the midline. (**e**) Closure of vaginal mucosa incision

in the transanal group as opposed to 93 % (14/15) in the transvaginal group. The need to digitate decreased significantly in both groups, from 93 % (14/15) to 27 % (4/15) in the transanal group and from 73 % (11/15) to 7 % (I/15) in the transvaginal group. Clinically diagnosed recurrence rates, however, showed a significant difference with 40 % (6/15) in the transanal group and 7 % (1/15) in the transvaginal group ($p=0.04$). There were no adverse effects on sexual function in either group, but 27 % (8/30) reported improvement.

In terms of functional results, the endoanal and transanal approaches appear similar, but data regarding long-term follow-up are largely unavailable. The use of mesh implantation again suffers from short follow-up times and/or small numbers, with the pervading worry of prosthetic material

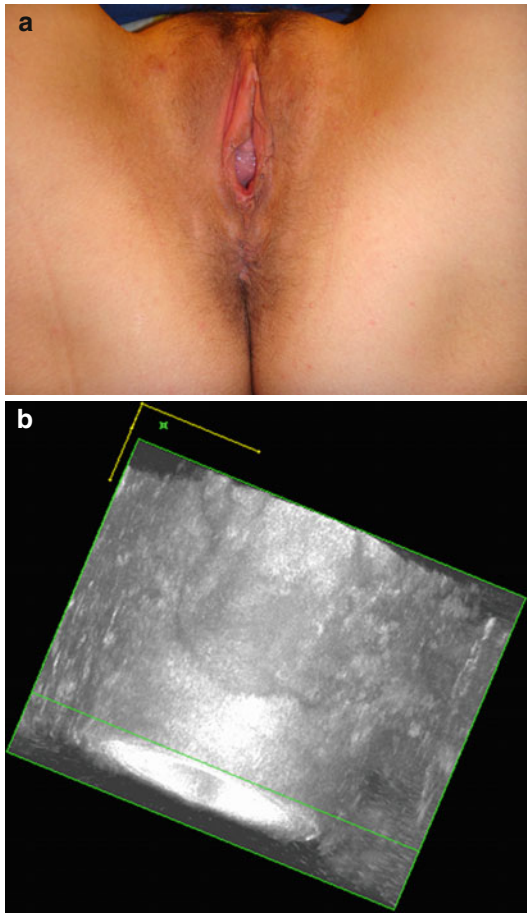

Fig. 9.5 (**a**) Damage of the left puborectalis with deviation of the anus on the healthy side. (**b**) Damage of the left puborectalis on sonography

erosion or infection and de novo dyspareunia. The clinical use of prosthetic materials in this area remains unknown, and there is a need for further study [44–46].

Type III Rectocele Associated with Invagination and/or Prolapse of the Rectum

Type *III* rectoceles are treated by an abdominal or a combined abdominoperineal approach. The abdominal part of the operation may be open or laparoscopic, and the procedure may include the placement of prosthetic material. The principle of the surgery is to perform a rectopexy to deal with the rectal prolapse; to repair the rectocele, through a perineorrhaphy; and to perform a Douglasorrhaphy in case of a deep pouch. This approach allows a correction of bladder or uterine prolapse, and in the case of a previous hysterectomy, the upper part of the vagina may be secured to the upper part of the levators or fixed to the sacrospinal ligament to prevent a vaginal vault prolapse. This abdominoperineal approach was described by Zacharin [28] in which the pouch of Douglas and the rectovaginal septum are opened. Marc Claude Marti added in 1992 to this technique a rectopexy to correct the rectal prolapse. Then, the pelvic hiatus is closed with

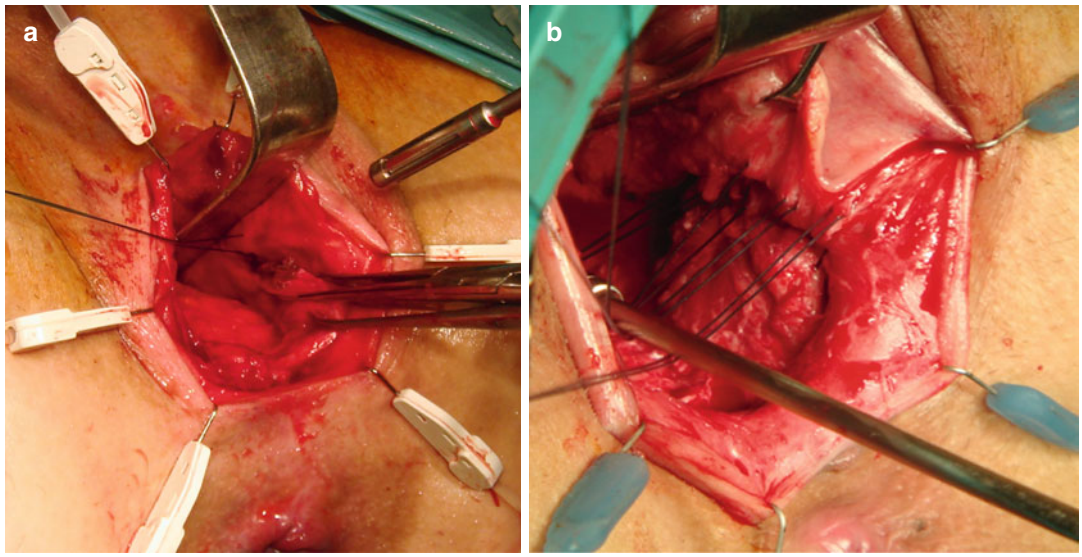

Fig. 9.6 Puborectalis repair on the left branch sutured to the pubis

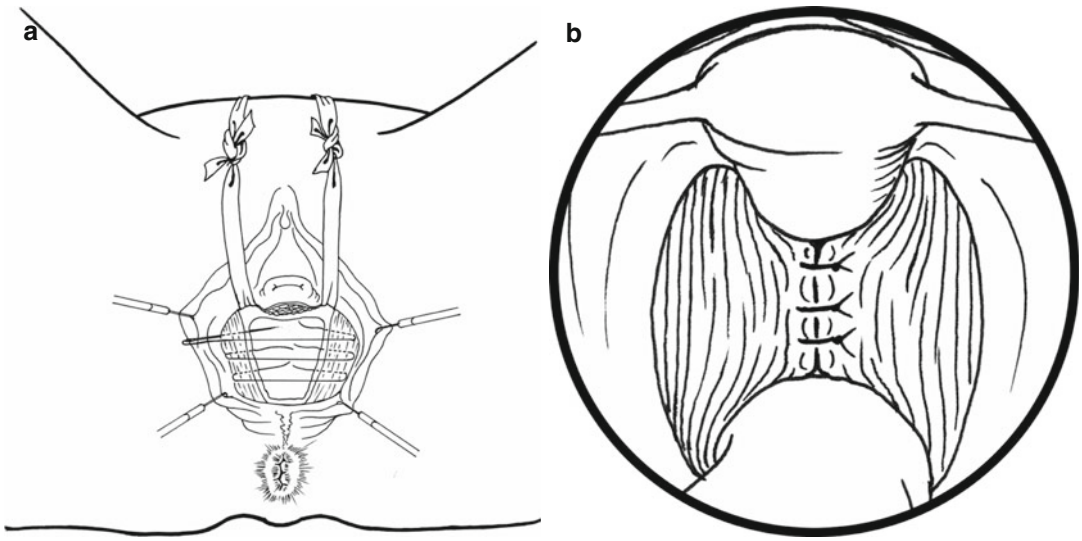

Fig. 9.7 Type III rectocele treatment according Marti-Zacharin procedure: (**a**) The pelvic hiatus is closed. (**b**) Laparoscopic aspect with three to four sutures placed through the levators on both sides

three to four sutures placed through the levators on both sides. The gauze strips are removed and a posterior colpomyorrhaphy is performed. We actually perform this procedure under laparoscopy (Fig. 9.7a, b).

Laparoscopic ventral mesh rectopexy is another technique described for rectal prolapse repair [29] that may be tailored to achieve Douglasorrhaphy and rectocele repair with or without perineal intervention. Fox and Stanton [45] describe mesh interposition to correct a rectocele at the time of abdominal sacrocolpopexy for vaginal vault prolapse. This simplifies the approach for patients with both vaginal vault prolapse and rectocele, as it alleviates the need for a concomitant vaginal procedure. The rectum is dissected from the posterior vaginal wall to the perineal body, and a continuous piece of mesh is placed from the perineal body to the vaginal vault. The mesh is then tied to the anterior longitudinal ligament overlying the sacral promontory in a tension-free fashion. The authors treated 29 patients with this surgery and reported significant improvement in prolapse symptoms. They were dissatisfied, however, by the continued bowel symptoms including constipation and incomplete defaecation. Similarly, Taylor et al. [46] reported a persistence or increase in bowel symptoms in 39 % of their patients who underwent this type of surgery.

Laparoscopic rectocele repair involves opening the rectovaginal space and dissecting inferiorly to the perineal body. The perineal body is sutured to the rectovaginal septum, and rectovaginal fascial defects are identified and closed. The levator ani muscles may be plicated. The advantages are reported to be better visualization secondary to magnification and insufflation and more rapid recovery, with decreased pain and hospitalization. Disadvantages are many, including difficulty with laparoscopic suturing, increased operating time and expense and an extended learning curve [50]. Moreover few reports describing outcomes of laparoscopic surgery for pelvic organ prolapse exist in the literature. Lyons and Winer [51] described the use of polyglactin mesh in laparoscopic rectocele repair in 20 patients, with 80 % reporting relief of both prolapse symptoms and the need for manual assistance to defaecate. Further studies are needed to assess this surgical approach for rectocele repair.

Conclusion

Rectoceles may be related to puborectalis muscle dysfunction. Overall, surgical correction success rates are quite high when using a

vaginal approach for rectocele correction. Vaginal dissection results in better visualization and access to the endopelvic fascia and levator musculature. This allows for a more solid and precise anatomical correction. Further studies and more comprehensive data collection are necessary to better understand the effect of various rectocele repair techniques on defaecatory, vaginal and sexual symptoms.

References

1. Dietz HP, Steensma AB. The role of childbirth in the aetiology of rectocele. BJOG. 2006;113:264–7.
2. Marks MM. The rectal side of the rectocele. Dis Colon Rectum. 1967;10:387–8.
3. Nichols DH. Sacrospinous fixation for massive eversion of the vagina. Am J Obstet Gynecol. 1982;142: 901–4.
4. Marti MC, Roche B, Deléaval J. Rectoceles: value of videodefaecography in selection of treatment policy. Colorectal Dis. 1999;1:324–9.
5. Shorvon PI, McHugh S, Diamant NE, et al. Defecography in normal volunteers: results and implications. Gut. 1989;30:1737–49.
6. Wald A, Caruana BI, Freimanis MG, et al. Contributions of evacuation proctography and anorectal manometry to evaluation of adults with constipation and defecatory difficulty. Dig Dis Sci. 1990;35: 481–7.
7. Yoshioka K, Matsui Y, Yamada O, et al. Physiologic and anatomic assessment of patients with rectocele. Dis Colon Rectum. 1991;34:704–8.
8. Halligan S, Bartram CL. Is barium trapping in rectoceles significant? Dis Colon Rectum. 1995;38: 764–8.
9. Stanton SL. Vaginal prolapse. In: Shaw RV, Souter WP, Stanton SL, editors. Gynaecology. Edinburgh: Churchill Livingstone; 2007. p. 764–6.
10. Block IR. Transrectal repair of rectocele using obliterative suture. Dis Colon Rectum. 1986;29:707–11.
11. Karlbom U, Graf W, Nilsson S, et al. Does surgical repair of a rectocele improve rectal emptying? Dis Colon Rectum. 1996;39:1296–302.
12. Mellgren A, Anzen B, Nilsson BY, et al. Results of rectocele repair. A prospective study. Dis Colon Rectum. 1995;38:7–13.
13. Khubchandani IT, Sheets JA, Stasik JJ, et al. Endorectal repair of rectocele. Dis Colon Rectum. 1983;26:792–6.
14. Sarles IC, Arnaud A, Selezneff I, et al. Endo-rectal repair of rectocele. Int J Colorectal Dis. 1989;4: 167–71.
15. Sehapayak S. Transrectal repair of rectocele: an extended armamentarium of colorectal surgeons. A report of 355 cases. Dis Colon Rectum. 1985;28:422–33.
16. Sullivan ES, Leaverton GH, Hardwick CE. Transrectal perineal repair: an adjunct to improved function after anorectal surgery. Dis Colon Rectum. 1968;11:106–14.
17. Altomare DF, Rinaldi M, Veglia A, et al. Combined perineal and endorectal repair of rectocele by circular stapler: a novel surgical technique. Dis Colon Rectum. 2002;45:1549–52.
18. Boccasanta P, Venturi M, Stuto A, et al. Stapled transanal rectal resection for outlet obstruction: a prospective, multicenter trial. Dis Colon Rectum. 2004;47: 1285–96.
19. D'Avolio M, Ferrara A, Chimenti C. Transanal rectocele repair using EndoGLA: short-term results of a prospective study. Tech Coloproctol. 2005;9:108–14.
20. Mathur P, Ng KH, Seow-Choen F. Stapled mucosectomy for rectocele repair: a preliminary report. Dis Colon Rectum. 2004;47:1978–80.
21. Bresler L, Rauch P, et al. Transrectal repair of rectocele using autosuture device. J Chir. 1993;130:304–8.
22. Maurel J, Gignoux M. Surgical treatment ofsupralevator rectocele. Value of transanal excision with automatic stapler and linear suture clips. Ann Chir. 1993; 47:326–30.
23. Regadas FS, Regadas SM, Rodrigues LV, et al. Transanal repair of rectocele and full rectal mucosectomy with one circular stapler: a novel surgical technique. Tech Coloproctol. 2005;9:63–6.
24. Lamah M, Ho J, Leicester RJ. Results of anterior levatorplasty for rectocele. Colorectal Dis. 2001;3:412–6.
25. Lehur PA, Bruley DV, Moyon J, et al. Disabling rectocele: rectal plication by perineal approach. A propos of 20 cases. Chirurgie. 1992;118:516–20.
26. Maeda K, Maruta M, Hanai T, et al. Transvaginal anterior levatorplasty with posterior colporrhaphy for symptomatic rectocele. Tech Coloproctol. 2003;7:181–5.
27. Nieminen K, Hiltunen KM, Laitinen J, et al. Transanal or vaginal approach to rectocele repair: a prospective. Randomized pilot study. Dis Colon Rectum. 2004;47: 1636–42.
28. Zacharin FR. Pelvic floor anatomy and the surgery of pulsion enterocele. New York: Springer; 1986. 170p.
29. D'Hoore A, Penninckx F. Laparoscopic ventral recto(colpo)pexy for rectal prolapse: surgical technique and outcome for 109 patients. Surg Endosc. 2006;20:1919–23.
30. Paraiso MF, Barber MD, Muir TW, et al. Rectocele repair: a randomized trial of three surgical techniques including graft augmentation. Am J Obstet Gynecol. 2006;195:1762–71.
31. Sarles IC, Ninou S, Arnaud A. Rectoceles. Diagnosis and treatment. Chirurgie. 1991;117:618–23.
32. Murthy VK, OrlOn BA, Smith L, et al. Excellent outcome using selective criteria for rectocele repair. Dis Colon Rectum. 1996;39:374–8.
33. Liberman H, Hughes C, Dippolito A. Evaluation and outcome of the Delorme procedure in the treatment of rectal outlet obstruction. Dis Colon Rectum. 2000;43: 188–92.
34. Trompetto M, Clerico G, Realis LA, et al. Transanal Delorme procedure for treatment of rectocele associ-

ated with rectal intussusception. Tech Coloproctol. 2006;110:389.
35. Ellis CN. Stapled transanal rectal resection (STARR) for rectocele. J Gastrointest Surg. 2007;11:153–4.
36. Ommer A, Albrecht K, Wenger F, et al. Stapled transanal rectal resection (STARR): a new option in the treatment of obstructive defecation syndrome. Langenbecks Arch Surg. 2006;391:32–7.
37. Pechlivanides G, Tsiaoussis J, Athanasakis E, et al. Stapled transanal rectal resection (Starr) to reverse the anatomic disorders of pelvic floor dyssynergia. World J Surg. 2007;31:1329–35.
38. Dodi G, Pietroletti R, Milito G, et al. Bleeding, incontinence, pain and constipation after STARR transanal double stapling rectotomy for obstructed defecation. Tech Coloproctol. 2003;17:148–53.
39. Bassi R, Rademacher J, Savoia A. Rectovaginal fistula after STARR procedure complicated by haematoma of the posterior vaginal wall: report of a case. Tech Coloproctol. 2006;110:361–3.
40. Boccasanta P, Venturi M, Salamina G, et al. New trends in the surgical treatment of outlet obstruction: clinical and functional results of two novel transanal stapled techniques from a randomised controlled trial. Int J Colorectal Dis. 2004;19:359–69.
41. Parker MC, Phillips RK. Repair of rectocoele using Marlex mesh. Ann R Coll Surg Engl. 1993;75: 193–4.
42. Altman O, Mellgren A, Zetterstrom I. Rectocele repair using biomaterial augmentation: current documentation and clinical experience. Obstet Gynecol Surv. 2005;60:753–60.
43. Huebner M, Hsu Y, Fenner DE. The use of graft materials in vaginal pelvic floor surgery. Int J Gynaecol Obstet. 2006;92:279–88.
44. Lim YN, Muller R, Corstiaans A, et al. A long-term review of posterior colporrhaphy with Vypro 2 mesh. Int Urogynecol J Pelvic Floor Dysfunct. 2007;18:1053–7.
45. Fox SD, Stanton SL. Vault prolapse and rectocele: assessment of repair using sacrocolpopexy with mesh interposition. Br J Obstet Gynaecol. 2000;107: 1371–5.
46. Taylor GM, Ballard P, Jarvis GJ. Vault prolapse and rectocele: assessment of repair using sacrocolpopexy with mesh interposition. Br J Obstet Gynaecol. 2001;8:775–6.
47. Backer Jr MH. Success with sacrospinous suspension of the prolapsed vaginal vault. Surg Gynecol Obstet. 1992;175:419–20.
48. Imparato E, Aspesi G, Rovetta E, et al. Surgical management and prevention of vaginal vault prolapse. Surg Gynecol Obstet. 1992;175:233–7.
49. Beck D, Allen N. Rectocele. Clin Colon Rectal Surg. 2010;23:90–8.
50. Paraiso MFR, Falcone T, Walters MD. Laparoscopic surgery for enterocele, vaginal apex prolapse and rectocele. Int Urogynecol J. 1997;8:146–52.
51. Lyons TL, Winer WK. Laparoscopic rectocele repair using polyglactin mesh. J Am Assoc Gynecol Laparosc. 1997;4:381–4.

Incontinence: Causes and Pathophysiology

10

Jonathan Randall and Ridzuan Farouk

Introduction

The rectum and anus, as the terminal segment of the gastrointestinal tract, have the primary function of storage and expulsion of faeces and flatus. With education this function is modified so that expulsion is reserved until the desire to expel rectal contents is sufficiently strong and social circumstances are appropriate.

Quality of life in patients with faecal incontinence as measured by the Faecal Incontinence Severity Index and Faecal Incontinence Quality of Life Scale is generally poorer than that of a 'normal' population when assessing social functioning and mental health. Faecal incontinence, when defined as the uncontrolled loss of solid or liquid stool, is difficult to quantify as not all patients will seek medical attention but is thought to have a prevalence of between 1 and 10 %. This chapter will consider the reasons why continence mechanisms break down. Firstly, consideration will be given to the major theories of continence, followed by a discussion of all the factors that usually contribute to continence and where they can fail, leading to incontinence.

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Theories of Continence

A number of different theories have been proposed to explain anorectal continence.

The Flutter Valve Theory

Following similar principles to mechanisms of gastro-oesophageal continence to prevent acid reflux, this theory tries to fit a mechanical explanation to continence [1]. In this theory it is supposed that raised intra-abdominal pressure occludes the high-pressure lower rectum, preventing movement of rectal contents into the anal canal. In this theory it was supposed that the sphincters were limited to fine control of continence and against sudden increases in rectal pressure. However, many studies have not found supporting evidence for this theory; for instance, Duthie [2] found that the mid-portion of the anal canal was the only zone that had a consistently high pressure.

The Flap-Valve Theory

Popularised by Sir Alan Parks [3], this theory developed the mechanical concept of continence but suggested that it was due to intra-abdominal forces acting against an acute resting anorectal angle. The puborectalis muscle acts like a sling and helps maintain the angle, and as the intra-abdominal pressure rises, so the anterior rectal wall 'plugs' the proximal anus. Some studies using videoproctography

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have not confirmed this, showing no contact between the anterior rectal wall and anal canal during Valsalva [4]. Indeed recent studies have shown little difference in the anorectal angle between incontinent patients and a normal group.

In Parks' theory a too obtuse angle, associated with perineal descent, would result in incontinence. To correct this, the postanal repair was developed. However, improvement in continence following this procedure has not been associated with an increased acuity in the anorectal angle [5].

Sphincteric Theories

Sphincteric theories of continence explain the effects of damage to the anal sphincters on continence, which can now be better demonstrated by endoanal ultrasound and MRI. The most important mechanism at rest is thought to be the internal anal sphincter (IAS) that forms a high-pressure zone at approximately 2 cm from the anal verge thus preventing entry of rectal contents. The internal anal sphincter is under a combination of controls, most coming from myogenic tone but with some augmentation from sympathetic nerves. Parasympathetic nerves are inhibitory. The IAS alone is unable to maintain complete occlusion, and the canal sits as an anteroposterior slit. Anal endocushions may generate up to 15 % of resting anal pressures and contribute to this occlusion [6].

The external sphincters are more important for generating additional pressure at times of threatened continence. Bartolo [4] found that continence was maintained in subjects in whom their rectum was filled with liquid by recruiting the external anal sphincter and puborectalis muscle. When a subject is asked to voluntarily contract their anal sphincters, the canal pressures are seen to rise by 175–270 %. The distribution of the contraction is symmetrical apart from the anterior where the pressures are significantly lower [7].

Breakdown in Normal Continence

Normal continence is dependent on a complex interaction between local and reflex mechanisms and voluntary intervention. Colonic transit and the consistency of stool being passed are important additional integral factors to maintenance of continence. The rectum is able to act as a reservoir, increasing in volume until a threshold is reached when the subject feels a desire to defecate. The normal subject can exhibit control over when it is appropriate to defecate, as well as discriminate between gas, fluid and solid faeces. Any breakdown in this system can lead to varying degrees of incontinence. It is important therefore to examine in detail the normal processes of rectal filling, sensation and defecation to understand the causes of incontinence.

Rate and Consistency of Delivery of Stool

Rapid rectal filling of very liquid stool can result in urgency and sometimes overcome the normal processes of continence. Any underlying sphincteric weakness may make individuals prone to leakage when stools are too liquid. Any cause of diarrhoea needs to be established before looking too closely for an underlying anatomical cause in the anorectum.

The rate of delivery of stool to the rectum may influence its ability to act as a reservoir through compliance (see below). The angle of the rectosigmoid junction, the resistance provided by the valves of Houston and a reduction in frequency of contractions between the sigmoid and rectum may all play a part in controlling the rate of delivery of stool to the rectum. Continence may be impaired when the rectosigmoid is diseased, as in inflammatory colitis, reducing the control of flow into and from the rectum. Excision of any part of the left colon may also speed up the transit of contents into the lower rectum, leading to urgency.

Rectal Compliance

Abnormal rectal function has been found in patients with faecal incontinence. Patients with normal anal pressure who are incontinent have been demonstrated to have abnormal rectal contractility [8]. Rectal compliance measures the relationship between changes in rectal volume with pressure. Compliance is the change in volume or cross-sectional area divided by the

change in pressure. Pressure/volume measurement during distention with a compliant balloon is the most commonly used method for computation of rectal compliance. However, intraindividual and interindividual variations exist. Other methods exist such as rectal distention by a large, noncompliant bag and rectal impedance planimetry for assessment of pressure-cross-sectional-area relations. This has been proposed as an alternative owing to the reduction of errors from elongation of the balloon within the rectal lumen. The rectum should be able to accommodate a significant volume without a significant rise in pressure. Interpretation of rectal compliance in the literature should consider the technique used [9].

Compliance can change with age but is also altered by disease states. Compliance is reduced in ulcerative colitis, Crohn's disease and radiation proctitis. In such states there can be an increased tendency to incontinence. In other forms of incontinence, it is possible the reduction in rectal compliance seen is secondary rather than a primary phenomenon.

Anorectal Sensation

The anal canal is sensitive to touch, pain, temperature and movement [10]. This aids continence through discrimination between fluid, flatus and faeces. Temperature discrimination is thought to be particularly important to the discrimination of solid stool from liquid stool and flatus. Temperature and electrical sensations are reduced in those with faecal incontinence [11], as well as with haemorrhoids and diabetic neuropathy.

Patients with rectal prolapse often suffer incontinence that is not associated with a desire to defecate, and soiling may only be discovered on inspection of underclothes. This may be attributed to the relatively insensitive rectal mucosa prolapsing into the anus. Furthermore, an improvement in anal continence status is associated with an improvement in anal sensation following anterior sphincter repair [12].

Patients who have undergone complete rectal resection and coloanal anastomosis continue to have some sensation, which improves over time, indicating that sensory receptors must also lie in the pelvic floor fascia or musculature [13].

Recto-anal Inhibitory Complex and Sampling

The anorectal inhibitory reflex occurs when there is a short-lived relaxation of the internal anal sphincter in response to rectal distension. This causes a transient reduction in resting anal pressures. Faecal matter or flatus then comes into contact with specialised anal receptors to allow sampling and discrimination of the state of the rectal contents to allow controlled passage of flatus without soiling. Crucially, the external sphincter is recruited during sampling to ensure continence. If defecation is not appropriate, the pelvic floor muscles return the contents upwards. The frequency of sampling reflexes varies depending on how it is measured (see below), but ambulatory manometry has suggested normal individuals have a rate of about 4 reflexes per hour that is increased following meals and prior to defecation [14]. These increases may be related to daytime rectal motor complex activity. In this study about one-third of events were not consciously perceived as a desire to defecate.

The rectal receptors responsible for this reflex are blocked by topical anaesthesia, which suggests they are located in the mucosa. Distension results in inhibition of the internal anal sphincter muscle fibres, mediated by nitric oxide. The role of the extrinsic autonomic system in this reflex is less clear. Anal relaxation may be absent in progressive rectal distension with low infusion rates, suggesting the internal anal sphincter can maintain continence when the rectum is slowly filling.

The sampling reflex has been demonstrated in the laboratory with rectal insufflation with a balloon which results in IAS relaxation. Ambulatory manometry [14] has shown frequent IAS relaxations with equalisation of rectal and upper anal canal pressures. Whether transient internal sphincter relaxations are initiated by the movement of rectal contents or are centrally mediated is still unclear.

Transient sphincter relaxation is abnormal in some faecally incontinent patients. The rise in rectal pressure and fall in mid-anal canal pressure is of greater magnitude than normal controls. The frequency of such relaxations is significantly higher in incontinent groups. This is perhaps a reflection of the abnormal internal sphincter in such patients as

evidenced by low resting pressures [8] as well as EMG abnormalities [14, 15] and increased collagen content [16]. The fall in mid-anal canal pressures may also reflect poor external sphincter recruitment in incontinent patients, particularly those with obstetric injuries who are aware of impending leakage but are unable to recruit sufficient external sphincter to prevent this happening.

Full-thickness rectal prolapse is associated with incontinence in 35–100 % of cases. Improvement in continence may be seen following rectopexy in combination with an improvement in anal sensation and resting anal pressure. It is plausible that the prolapse, which starts as an intussusception of the upper rectum, acts like a faecal bolus and induces an anorectal inhibitory reflex which results in internal sphincter relaxation and a fall in anal pressures. In fact studies show that eventually the reflex may not be elicited, perhaps because the internal anal sphincter is maximally inhibited [15]. In addition, patients with prolapse exhibit a different type of high-pressure rectal wave that results in prolonged anorectal inhibition and fall in anal pressures. They are recorded in patients with complete rectal prolapse [15] where anal pressures fall to levels that would promote faecal leakage.

The anorectal inhibitory reflex is not seen to occur in Hirschsprung's disease where the myenteric plexus is absent in the rectum. After rectal excisional surgery and coloanal anastomosis, the reflex is initially absent but it rapidly recovers.

Nerve Supply

The motor innervation of the rectum and anal canal is classified into its intrinsic and extrinsic motor supply. The intrinsic nerve supply is via the myenteric and submucosal plexus with ganglia that are interconnected by numerous nerve fibres. The autonomic and somatic nervous systems have to function in an integrated manner. The sympathetic neurons originate as cell bodies in the lumbar spinal cord (L2 to L4). The parasympathetic nerve supply of the anorectum has its origins in the sacral segments of the spinal cord (S2 to S4). The cell bodies of the somatic nervous innervation of the external anal sphincter and the striated muscles lie in the ventral horn of the sacral spinal cord (S1 to S3). The external sphincter is supplied by the pudendal nerve, and the posterior sphincter receives a direct branch from the perineal branch of S4. The levator ani group of muscles is innervated by the third and fourth sacral nerves and the pudendal nerve (S2, S3, S4).

Pelvic floor denervation can occur with any cause of neuropathy including diabetes, trauma and malignant infiltration. It has been suggested that excessive pelvic floor descent causes traction and damage to the pudendal nerve leading to muscular atrophy [17]. This has been linked to faecal incontinence amongst other conditions. Kiff and Swash [18] showed slowed conduction in the pudendal nerves in idiopathic (neurogenic) faecal incontinence. Measured by the St Marks' perineometer, pelvic floor descent can cause significant stretching and lead to permanent damage [19]. Irreversible damage occurs after a stretch of 12 % of the pudendal nerve. Although a relationship has been seen between pelvic floor descent and pudendal nerve terminal motor latency in patients with faecal incontinence [20], the degree of incontinence seen does not correlate with the extent of pelvic floor descent [21].

Intact Sphincters

The internal sphincter is an involuntary muscle that is a continuation of the circular muscle coat of the rectum. Surrounding the upper two-thirds of the anal canal, it is approximately 3–4 cm long and varies in thickness between 0.1 and 0.5 cm. It is shorter in women and shaped like a signet ring. Higher mean resting pressures are seen in men and decline in old age, particularly in women. Resting anal pressure undergoes regular fluctuations with an amplitude of 5–25 cm H_2O and a frequency of 10–20/min. These slow waves are thought to be generated by the internal sphincter [22]. The frequency of the slow wave is higher in the lower anal canal providing a mechanism which returns contents upwards. In addition, a zone of high pressure exists approximately 2 cm from the anal verge which is longer in men than women.

The contribution of the internal anal sphincter to resting pressures, which is thought to be around 85 %, has been established by pudendal nerve blocks and as seen after division of the internal sphincter.

The external anal sphincter is striated and traditionally divided into subcutaneous, superficial and deep layers [23]. It is composed of both slow- and fast-twitch fibres. Reflex contraction should occur on coughing or any rise in intra-abdominal pressure. The external anal sphincter and puborectalis generate maximal squeeze pressure, usually an additional pressure of between 50 and 200 mmHg.

The puborectalis muscle forms a part of the pelvic floor musculature and exhibits almost identical electrophysiological characteristics with the external anal sphincter during coughing and straining.

In general, internal anal sphincter pathology can lead to passive anal leakage whilst stress or urge incontinence may be attributed to problems with the external anal sphincter. In patients with incontinence, both resting and maximal squeeze pressures are lower than matched controls [24]. Common causes of sphincter disturbance include:

Obstetric Injuries

A higher proportion of women sustain sphincter damage during childbirth than what is suspected by immediate clinical assessment [25]. Primary repair of third-degree tears can be performed via an end to end or overlapping technique and can be very successful although infection or haematoma can lead to wound breakdown. Despite repair defects are often still visible on endoanal ultrasound scan and many women continue to have some degree of incontinence [26].

Patients with obstetric injuries and resulting faecal incontinence are often aware of impending leakage but are not able to recruit sufficient external sphincter to prevent this from happening. Leakage may occur during the previously described internal sphincter relaxation during sampling when recruitment of the external sphincter is poor. This can result in urgency and leakage.

Endoanal ultrasound and MRI are both frequently used to assess sphincter defects. Endocoil MRI may be better than ultrasound at assessing the extent of external sphincter defects and can also distinguish sphincter atrophy where the sphincter is replaced by fat and fibrous tissue [27].

Surgery for Anal Fissure

Anal fissures are frequently associated with high resting anal pressures. Historical treatments included anal stretch, a procedure that has since been shown to cause widespread sphincter damage [28] and has largely been abandoned. Although temporary sphincter paralysis can be used as a treatment with calcium antagonists or injection with botulinum toxin, resistant fissures may still be treated by intentional division of the internal sphincter. This results in lower resting tones, but in some cases, particularly where there has been previous sphincter injury, a degree of incontinence may ensue. Laterally placed sphincterotomies away from the fissure itself are recommended to avoid key hole deformities which have a higher incidence of loss of continence.

Surgery for Anal Fistulas

The most successful treatment of anal fistulas involves laying the tract open. Unfortunately even partial division of the sphincters can result in a degree of incontinence. Clinical outcomes correlate with the amount of sphincter cut as well as with the decrease in resting and squeeze pressures [29].

Haemorrhoid Surgery

A haemorrhoidectomy may be associated with damage to the internal sphincter if dissection occurs in the wrong plane [15]. Anal stenosis can also occur if insufficient tissue bridges are left between resected haemorrhoidal cushions leading to difficult evacuation and poor control with leakage.

Assessment of the Patient with Incontinence

History

The history should include making the diagnosis of incontinence, making an assessment of severity and consideration of aetiology.

Diagnosis

Patients may report a combination of incontinence episodes to flatus, liquid or solid stools. Patients should be questioned whether episodes are predominantly passive, with leakage and soiling or urge incontinence, particularly on coughing or straining.

Assessment of Severity

Frequency of incontinence episodes and to what (flatus, liquid or solid stool) can provide an index of severity. Effect on quality of life should be made by questions about hygiene and use of protective pads as well as restrictions on activities. A variety of incontinence scores have been developed to quantify the severity of incontinence. The Cleveland Clinic Incontinence Score assesses incontinence to solids, liquids and flatus as well as use of pads and lifestyle alteration.

Aetiology

The history should investigate potential causes of incontinence including:

- Obstetric history. Known injuries as well as questioning about number of difficult or assisted births may reveal a likely occult cause of incontinence. Obstetric injuries can cause both sphincter damage and nerve damage by impingement and stretch. Routine episiotomies do not reduce the incidence of incontinence although episiotomies for complicated deliveries may reduce the risk of anal sphincter damage.
- Previous anorectal surgery. As mentioned in section "Breakdown in normal continence", anorectal procedures including haemorrhoidectomy and surgery for fissure and fistula can lead to a degree of incontinence. In addition rectal resections and anastomosis can affect continence through loss of reservoir function and sensory mechanisms. However, as long as sphincter function is preserved, recovery of continence can usually be expected over a period of months.
- History of pelvic radiotherapy for treatment of prostatic, anal, rectal or gynaecological cancers can lead to radiation proctitis and, less commonly, sphincter damage. Radiation proctitis can lead to loss of rectal compliance and urgency and may be indicated by a history of rectal bleeding.
- Underlying colorectal pathology. As discussed in section "Breakdown in normal continence", conditions such as inflammatory bowel disease may lead to rapid transit and loss of rectal capacity that overwhelm normal continence. Questioning should also be directed towards an underlying neoplasm of the anus or rectum. A history of bleeding, diarrhoea and weight loss as well as family history should be sought.
- A history of straining and urinary incontinence favours pelvic floor pathology, usually neuropathic incontinence. A history of a prolapse that has to be reduced is associated with incontinence in over 50 % of cases. The association between prolapse and incontinence may be due to changes in sensation and local reflexes as discussed in section "Breakdown in normal continence".
- Trauma. Blunt or penetrating injuries including impalement can lead to both nerve injury and direct sphincter injury.

Examination

Features of the examination that will be useful to the diagnosis of incontinence include:

- Inspection: Soiling and use of pads may confirm incontinence as a presenting problem. Rectal prolapse, prolapsing haemorrhoids and scarring may point towards a cause. It is important to part the buttocks to closely examine the anal verge.
- Digital rectal examination: Resting and squeeze pressure can be examined with the finger as well as any defects in the sphincter.
- Anal sensation can be assessed by light touch and/or pinprick. Absence of the anal wink reflex may reflect underlying nerve damage.
- Direct visualisation of the anorectum with a proctoscope and rigid sigmoidoscope should be performed in clinic to view the anal and rectal mucosa.

Investigations

Endoanal Ultrasound Scans

The internal sphincter appears as a hypoechoic inner circle whilst the external sphincter appears as a hyperechoic structure. This allows sphincter defects and scarring to be visualised and described in relation to points on the clock face. Originally developed as a two-dimensional imaging technique, three-dimensional endoanal ultrasound is now available.

MRI

Endocoil receiver MRI may give better visualisation of external anal sphincter defects than endoanal ultrasound scans and can distinguish between true sphincter defects and atrophy.

Manometry

Using a variety of methods including water-filled perfusion catheters, water- or air-filled balloons, sleeve catheters and pressure transducers, manometry can be used to determine resting and squeeze pressures as well as mapping out the length and pressures of the anal canal. Both resting and squeeze pressures have been noted to be significantly lower in patients with incontinence compared to matched controls [24]. Ambulatory manometry allows an assessment of canal pressures in more physiological conditions.

Electromyography

The electrophysiology of the external sphincter and puborectalis muscle can be investigated by electromyography. Needle electrodes can pick up muscular fibrillation suggestive of nerve injury.

Pudendal nerve stimulation transanally provides a method of assessing pelvic floor neuropathy. Measurement of the latent period between pudendal nerve stimulation and electromechanical response of the muscle is termed 'pudendal nerve terminal motor latency'. It is prolonged in patients with idiopathic incontinence, prolapse and in certain neurological conditions [30].

Rectal Compliance

Pressure/volume curves are generated using water-filled balloons with internal sensors in the rectum. Values can be recorded for initial sensation to defecate, urgency and maximum tolerated volume.

Defecating Proctogram

If the rectum is filled with radio-opaque contrast and fluoroscopic images taken during defecation, then a dynamic recording can be made. Changes important in diagnosis of incontinence include perineal descent, prolapse, enterocoele, rectocele and changes in the anorectal angle. MRI defecography is an alternative imaging modality for individuals with incontinence to visualise anatomical changes during evacuation.

References

1. Phillips SF, Edwards DAW. Some aspects of anal continence and defecation. Gut. 1965;6:396–405.
2. Duthie H. Anal incontinence. Gut. 1971;12:844–52.
3. Parks A. Anorectal incontinence. Proc R Soc Med. 1975;68:681–90.
4. Bartolo D, Roe A, Locke-Edmunds J, et al. Flap-valve theory of ano-rectal incontinence. Br J Surg. 1986;73:1012–4.
5. Miller R, Bartolo DCC, Locke-Edmunds JC, Mortensen NJ. Prospective study of conservative and operative treatments for faecal incontinence. Br J Surg. 1988;75:101–5.
6. Lestar B, Penninckx F, Kerremans R. The internal anal sphincter cannot close the anal canal completely. Int J Colorectal Dis. 1992;7:159–61.
7. Taylor BM, Beart RW, Phillips SE. Longitudinal and radial variations of pressure in the human anal canal. Gastroenterology. 1984;86:993–7.
8. Sun WM, Read NW, Miner PB, Kerrigan DD, Donnelly TC. The role of transient internal sphincter relaxation in faecal incontinence. Int J Colorectal Dis. 1991;5:31–6.
9. Krogh K, Ryhammer AM, Lundby L, Gregersen H, Laurberg TS. Comparison of methods used for measurement of rectal compliance. Dis Colon Rectum. 2001;44:199–206.
10. Duthie H, Gairns F. Sensory nerve endings and sensation in the anal region of the man. Br J Surg. 1960;47:585–95.
11. Roe A, Bartolo D, Mortensen N. New method for assessment of anal sensation in various anorectal disorders. Br J Surg. 1986;73:310–2.
12. Orrom WJ, Miller R, Cornes H, Duthie GS, Mortensen NJ, Bartolo DCC. Comparison of anterior sphincteroplasty and postanal repair in the treatment of idiopathic faecal incontinence. Dis Colon Rectum. 1991;34:305–10.

13. Lane R, Parks A. Function of the anal sphincters following colo-anal anastomosis. Br J Surg. 1977;64: 596–9.
14. Farouk R, Duthie GS, Pryde A, McGregor AB, Bartolo DC. Internal anal sphincter dysfunction in neurogenic faecal incontinence. Br J Surg. 1993;80: 259–61.
15. Farouk R, Duthie GS, MacGregor AB, Bartolo DC. Evidence of electromechanical dissociation of the internal anal sphincter in idiopathic fecal incontinence. Dis Colon Rectum. 1994;37:595–601.
16. Swash M, Gray A, Lubowski D, Nicholls R. Ultrastructural changes in internal anal sphincter in neurogenic faecal incontinence. Gut. 1988;29:1692–8.
17. Swash M, Snook S, Henry M. Unifying concepts of pelvic floor disorders and incontinence. J R Soc Med 1985;78(11):906–11.
18. Kiff E, Swash M. Slowed conduction in the pudendal nerves in idiopathic (neurogenic) faecal incontinence. Br J Surg. 1984;71:614–6.
19. Schizas A, Williams A. Anorectal investigation. In: Phillips R, editor. Colorectal surgery. 4th ed. Edinburgh: Saunders/Elsevier; 2009.
20. Laurberg S, Swash M, Snooks S, Henry M. Neurologic cause of idiopathic incontinence. Arch Neurol. 1988; 45:1250–3.
21. Read N, Bartolo D, Read M, et al. Differences in anorectal manometry between patients with haemorrhoids and patients with descending perineum syndrome: implications for management. Br J Surg. 1983;70(11): 656–9.
22. Kerremans R. Morphological and physiological aspects of anal continence and defaecation. Brussels: Arscia; 1969.
23. Milligan M, Morgan C, Jones H, Officer R. Surgical Anatomy of the anal canal, and the operative treatment of haemorrhoids. The Lancet 1937;1150–6.
24. Bharucha A, Fletcher J, Harper C, et al. Relationship between symptoms and disordered continence mechanisms in women with idiopathic faecal incontinence. Gut. 2005;54:546–55.
25. Andrews V, Sultan AH, Thakar R, Jones PW. Occult anal sphincter injuries – myth or reality? BJOG. 2006; 113:195–200.
26. Sakse A, Secher NJ, Ottesen M, Starck M. Defects on endoanal ultrasound and anal incontinence after primary repair of fourth-degree anal sphincter rupture: a study of the anal sphincter complex and puborectal muscle. Ultrasound Obstet Gynecol. 2009;34(6):693–8.
27. Williams A, Bartram C, Modhwadia D, et al. Endocoil MRI quantifications of external anal sphincter atrophy. Br J Surg. 2001;88:8539.
28. Speakman CT, Burnett SJ, Kamm MA, Bartram CI. Sphincter injury after anal dilatation demonstrated by anal endosonography. Br J Surg. 1991;78:1429–30.
29. Sanino P. A manometric study of anorectal function after surgery for anal fistula, with special reference to incontinence. Acta Chir Scand. 1985;151:695–700.
30. Snooks SJ, Barnes RPH, Swash M. Damage to the voluntary anal and urinary sphincter musculature in childbirth. J Neurol Neurosurg Psychiatry. 1984;47: 1269–73.

Incontinence Assessment: Physiology and Imaging

11

Mostafa Abdel-Halim

Introduction

Assessment of faecal incontinence is a multifaceted process, which involves clinical assessment together with the appropriate physiological and imaging tests of the anorectum. The purposes of the process of evaluation of the patient are firstly, to assess the severity of the problem and its impact on the quality of life and, secondly, to identify as much as possible the underlying aetiological factors. Acquiring this knowledge enables the physician to design appropriate treatment strategies which could address the problem.

There is a big armamentarium of physiological and imaging investigations; and multiple tests are usually indicated in the process of evaluating the patient as aetiology is often multifactorial. Altered sphincter or pelvic floor mechanism, neuropathy, impaired anorectal sensations and inadequate rectal reservoir function may all contribute to the pathogenesis of faecal incontinence (Table 11.1 shows a list of causes of faecal incontinence). It is to be mentioned, however, that a careful clinical assessment remains essential and probably the most important element for an accurate evaluation.

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Clinical Assessment

History Taking

Careful clinical history is extremely important. It is often required to tease out the complaints voiced by the patient and establish the presence of the problem in the first instance, as often patients do not verbalise their symptoms directly. In such circumstances, the distressing problem of incontinence might remain unaddressed or be mistreated, if the physician omits detailed clear history taking and enquiry.

The nature of the incontinence and whether it is predominantly an urge or passive leakage sheds light on the potential underlying pathophysiology. Passive incontinence, which is the involuntary loss of faecal matter or flatus to variable extent without conscious awareness, suggests a loss of perception and/or impaired rectoanal reflexes either with or without sphincter dysfunction [1]. Urge incontinence, which is the loss of faecal matter or flatus to variable extent in spite of an active attempt to retain them, suggests predominantly a disruption of the sphincter function or the rectal capacity to function as a reservoir [1]. Detailed history will also allow the differentiation of perineal soiling or mucus staining which can result from the presence of haemorrhoidal or rectal mucosal prolapse, fistula-in-ano or perineal sexually transmitted lesions. Enquiry about the ability to discriminate between formed or unformed stools and gas should be made. Moreover, it is important to delineate any symptoms of difficulty in evacuation or pelvic organ prolapse.

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Table 11.1 Causes of faecal incontinence

A. Altered stool consistency (diarrhoeal states)
1. Inflammatory bowel diseases
2. Infectious diarrhoea
3. Malabsorption syndromes
4. Short gut syndrome
5. Laxative abuse
6. Irritable bowel syndrome
B. Inadequate rectal reservoir or compliance
1. Inflammatory bowel disease
2. Radiation enteritis
3. Surgical resection of the reservoir:
(a) Low anterior resection of the rectum
(b) Ileoanal pouch surgery
4. Rectal ischaemia
5. Rectal neoplasia
6. Extrinsic rectal compression
7. Scleroderma and other collagen disorders
C. Altered rectal sensory function or motility
1. Cerebrovascular stroke
2. Central neurological trauma or neoplasia
3. Multiple sclerosis
4. Tabes dorsalis
5. Spina bifida
6. Myelomeningocele
7. Dementia
8. Peripheral neuropathy
9. Impaired motility and resulting overflow incontinence:
(a) Faecal impaction
(b) Anti-motility drugs
(c) Psychotropic drugs
10. Rectal hypermotility syndrome
D. Altered sphincter or pelvic floor mechanism
I. Anatomical or mechanical deficit:
1. Obstetric sphincter injury
2. Iatrogenic injury post anorectal surgery:
(a) Anal fistula surgery (lay open procedures)
(b) Haemorrhoidectomy
(c) Sphincterotomy
(d) Dilatation or anal stretch
3. Trauma
4. Neoplasia involving the sphincter
5. Congenital defects of the sphincter and pelvic floor:
(a) Imperforate anus
(b) Anal agenesis
II. Neurological deficit:
1. Neurogenic incontinence:
(a) Pudendal neuropathy
(b) Post vaginal delivery
2. Injury to spinal cord (cauda equina) or pelvic floor nerves (e.g. pelvic surgery)
3. Spina bifida and myelomeningocele
4. Diabetic neuropathy
III. Functional deficit:
1. Ageing
2. Sphincter atrophy
3. Prolonged rectal prolapse
4. Increased body mass index

History of any precipitating factors, coexisting conditions such as diabetes mellitus or neurological conditions, past medical history, previous anorectal surgery, drug history and detailed obstetric history in female patients are also of great importance. An essential part of the history taking is establishing the extent of disruption and affliction on the patient's quality of life and the ability to pursue their daily activities and their degree of willingness to undergo an invasive intervention to address the problem. This will help direct treatment strategies which are primarily aimed at improving patients' quality of life and coping mechanisms.

Symptom Scores and Diaries

Validated incontinence scoring systems are an integral part of clinical assessment. Coupled with the use of prospective symptom diaries which supplement the snap shot questionnaire assessment by giving a record of clinical condition over the period of a week or more, they aid diagnosis, assessment of severity of symptoms and also help with monitoring the response to any treatment offered. A number of grading questionnaires have been designed aiming at quantifying the extent of symptoms into a numeric analogue assessment [2–4].

Examination

General and perineal examination is essential to identify the diagnosis. General examination may reveal a clue as to aetiology, such as signs of a neurological or connective tissue disorder. Inspection of the perianal area may reveal the

presence of skin excoriation from exposure to faecal matter, a patulous anus or perineal descent. Other abnormalities like haemorrhoidal or rectal mucosal prolapse may be also diagnosed. Perineal scarring and small or absent perineal body would suggest previous obstetric trauma. The perianal sensations and the anocutaneous reflex should be also checked as impaired sensations or absent reflex suggest neuronal injury.

Digital rectal examination enables the examiner to assess (a) the bulk of the sphincter, (b) the presence of palpable defects, (c) the tone of the sphincter, (d) the quality of the squeeze function, (e) the presence of any rectal masses or rectocele and (f) the observation of pelvic floor dyssynergia. Digital examination is also of value to detect faecal impaction which can be associated with overflow incontinence. If a patient is suspected of having a rectal prolapse, this needs to be further evaluated whilst the patient is straining in the seated position.

At this stage, it has to be distinguished whether the incontinence is secondary to diarrhoea and a change in bowel habit or independent of stool consistency. If there is evidence of a change in bowel habit or other warning symptoms or signs, endoscopy (and other tests accordingly) is indicated to rule out mucosal pathology or neoplasia. Discussion of such workup is not within the merits of this chapter. Table 11.2 summarises the main components of clinical assessment.

Physiological Studies

Several physiological tests which often complement each other are available to assess anorectal functions. Physiological tests provide an objective assessment of anal sphincter pressures, rectoanal reflexes, anorectal sensory function and rectal compliance.

Anal Manometry

Standard Anal Manometry

Technique and Measurements

The equipment required for this test consists of four components: a probe, a pressure recording device, record-displaying device and a data storage facility (Fig. 11.1). Two types of probes are used: a solid-state probe with strain gauge transducers and a water-perfused probe. Technique details and equipment configurations significantly vary and examples have been published by various groups [5, 6]. Manometry is used to assess the resting and squeeze anal pressures, the cough reflex and the rectoanal inhibitory reflex (RAIR) (Fig. 11.2). Rectoanal pressure changes during simulated defecation can be also assessed in certain cases.

Bowel preparation is not usually required but if digital rectal examination reveals faecal loading a preparatory rectal enema should be given. The patient is placed in the left lateral position with 90° flexion at the hips and knees. The procedure is preceded by digital rectal examination and the lubricated manometry probe is then gently inserted into the rectum. After a brief rest period to allow the sphincter tone to return to basal levels, measurements are taken during station pull through; the probe is gradually withdrawn in 0.5 or 1 cm steps and the recordings are taken when the transducer is stationery to avoid artefacts induced by reflex sphincter contraction during a rapid pull through procedure.

Resting anal pressure is defined as the difference between the intrarectal pressure and the maximum anal pressure at rest [5]. Pressures should be measured by averaging all four quadrants to account for sphincter asymmetry. Squeeze pressure is obtained by asking the patient to contract the sphincter for at least 30 seconds and to average the pressure measurement over this period [5]. The time interval in seconds during which the subject can maintain a squeeze pressure at or above 50 % of the maximum squeeze pressure represents the duration of sustained squeeze. The reflex increase in anal pressure during abrupt change in intra-abdominal pressure is tested by asking the patient to cough (cough reflex test) [7]. This protective reflex is thought to be mediated through a spinal reflex arc.

Clinical Utility

Studies have demonstrated high reproducibility of anal pressure measurements in the same subject on separate days [6]. Anal pressures are influenced

Table 11.2 Clinical assessment in faecal incontinence

History	
Personal history	
Onset, course and duration	
Nature of incontinence	Urge (sphincter or reservoir dysfunction)
	Passive (sensory dysfunction/internal sphincter dysfunction)
	Mixed
Ability to distinguish solid, liquid and flatus	
Prolapse symptoms	(Haemorrhoidal prolapse, rectal prolapse, global pelvic floor failure)
Frequency of bowel motions	(Very infrequent motions might suggest overflow)
Consistency of stools	(Diarrhoeal conditions and inflammatory bowel disease)
Difficulty in rectal evacuation	(Dyssynergia, global pelvic floor failure, neurological conditions)
Other bowel symptoms e.g. change in bowel habit, PR bleeding or tenesmus	(Investigate for mucosal pathology or neoplasia)
Past medical history	Diabetes mellitus
	Neurological conditions
History of previous surgery	(Incontinence related to previous anorectal surgery which damaged the sphincter)
Obstetric history	Number of pregnancies and deliveries
	Use of forceps
	Previous tears
Drug history	
Extent of interference with quality of life	
Examination	
General and back examination	(Signs of neurological disease or connective tissue disorder)
Inspection of the perianal area	Skin excoriation from faecal matter
	Haemorrhoidal Prolapse
	Scarring (Previous obstetric injury or anal surgery)
	Small or absent perineal body (Previous obstetric injury)
Test anocutaneous reflex (stroking the perianal skin with a cotton bud leads to a brisk contraction of the external sphincter)	(Loss of the reflex suggests neuronal injury. Afferents are sensory nerves to the skin. Centre is spinal segments S2, 3, 4. Efferents are motor supply to external sphincter)
Digital rectal examination	Is there faecal impaction
	Assess the bulk of the sphincter
	Assess the presence of any palpable defects
	Assess the tone of the sphincter
	Assess the squeeze of the sphincter
	Check for rectocele
	Assess for dyssynergia of the pelvic floor
Check for rectal prolapse if suspected (straining in the seated position)	

by age and gender [7–9]; men have higher mean resting and squeeze pressures [10, 11] and pressures decline after the age of 60 years [10, 12]. There is relative lack of age- and sex-stratified normative data, and it is suggested that individual centres establish their own [5, 8]. Moreover, consideration of the different methods used to obtain the pressure measurements is essential when comparing the results of different studies.

The resting anal pressure predominantly represents the internal anal sphincter function and the voluntary squeeze pressure predominantly represents the external sphincter function. Normally, the high pressure zone which is the part of the anal canal where the resting pressure is >50 % of the maximum resting pressure is longer in males than females [13, 14]. In a normal individual the rise in pressure on

Fig. 11.1 Manometry stack

maximal squeeze should be at least 50–100 % of the resting pressure [15].

Anal pressures are usually reduced in faecal incontinence [16–18]; however, in cases where aetiology does not involve sphincter malfunction (e.g. diarrhoea, disturbances of rectal sensations or compliance), anal pressures can be within normal range. Moreover, in a significant proportion of asymptomatic normal individuals, pressures were found to be low [19].

Differential reduction in anal pressures could reflect the site of deficit; a relatively maintained squeeze pressure with low resting pressure reflects a deficit in IAS function and vice versa. Reduced resting pressure is associated with passive leakage, whilst reduced squeeze pressure is associated with inability to delay rectal evacuation [20]. The loss of the normal cough reflex together with the voluntary squeeze function denotes a cauda equina lesion or a lesion in the sacral plexus, whereas in patients with spinal cord lesions above the conus medullaris, this

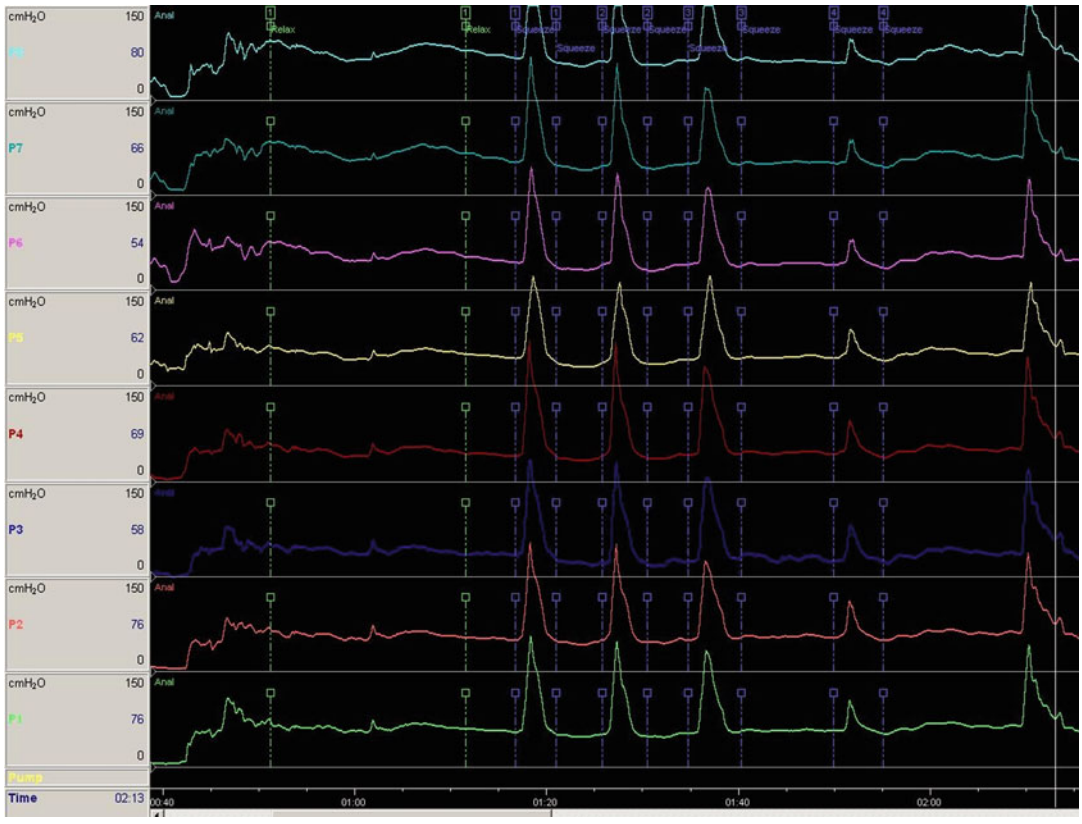

Fig. 11.2 Eight channel manometry trace (RAIR is not viewed on this trace)

reflex is present but the voluntary squeeze may be absent [21, 22].

Treatment of faecal incontinence cannot be monitored by changes in anal pressures. In some studies, anal pressures increased after biofeedback therapy; however, the magnitude of change was small and did not correlate with symptom improvement [23]. Similarly, changes in anal pressures following successful treatment with sacral nerve stimulation have been inconsistent [24].

Other Manometric Techniques

Ambulatory Manometry

This involves the use of micromanometers mounted on a catheter to simultaneously record rectal and anal canal pressures in a continuous manner [25, 26]. Although it provides information on the functioning sphincter in a more physiological situation in comparison with the static manometry, its use is primarily employed in the research setting.

Vector Volume Manometry (Vectometry)

This technique utilises a special device to automatically withdraw an eight-channel manometry catheter from the anal canal during rest and squeeze; and with the use of computer software, a three-dimensional reconstruction of the anal canal is then produced [27]. Vectrometry can be used to differentiate between global sphincter atrophy or weakness and traumatic sphincter injury by demonstrating global weakness or significant asymmetry indicating a localised sphincter abnormality respectively [27, 28]. However, poor correlation between the asymmetry on the vectogram and the electromyographic or ultrasound localisation of sphincter defects was demonstrated in some studies [29] and endoanal ultrasound remains the gold standard for identifying anal sphincter defects [30]. Nevertheless, a recent review suggests that vectrometry results correlate well with imaging and that clinical utility can be improved with the standardisation of technique and equipment [31].

High-Resolution Anorectal Manometry

This new approach utilises high-resolution techniques in combination with novel rapid data-interpretation software to allow the interpolation of manometric recordings into highly detailed topographical plots of the anorectal intraluminal pressure events relative to time [32]. Results of this technique seem to be well collaborated with conventional measurements and further studies are being conducted to establish its place in routine assessment.

Recto-Anal Inhibitory Reflex (RAIR)

Reflex relaxation of the upper internal anal sphincter (IAS) occurs with progressive rectal filling and is a physiological phenomenon vital to the continence mechanism. Representing the close association between anorectal sensations and motor function, this ‘sampling reflex’ occurs regularly allowing rectal contents to be presented to the specialised lower anal sensory mucosa where it can establish its nature (solids, fluid or gas) [33, 34]. The contents are delivered back to the rectum by the effect of the high pressure generated by the presence of the external anal sphincter (EAS) overlapping the lower anal canal [35]. Figure 11.3 demonstrates the various components of this reflex.

Technique and Measurement

The RAIR is recorded as part of the anorectal manometry study. This normal reflex is elicited by rapidly inflating a balloon in the rectum whilst simultaneously recording the anal pressure to demonstrate the IAS relaxation. Most currently available manometry catheters are manufactured with a mounted balloon to facilitate this test (Fig. 11.4).

Clinical Utility

The RAIR was first described by Gowers in 1877 [36] and was later confirmed in 1935 by Denny-Brown and Robertson [37]. It is thought to be mediated via the intramural neuronal plexus [34], and its absence is used as a diagnostic criterion for Hirschsprung’s disease [38–40]. The reflex is also absent following rectal resections; however, there is some evidence that it recovers with time [41, 42]. Some studies have demonstrated specific changes to this reflex in association with

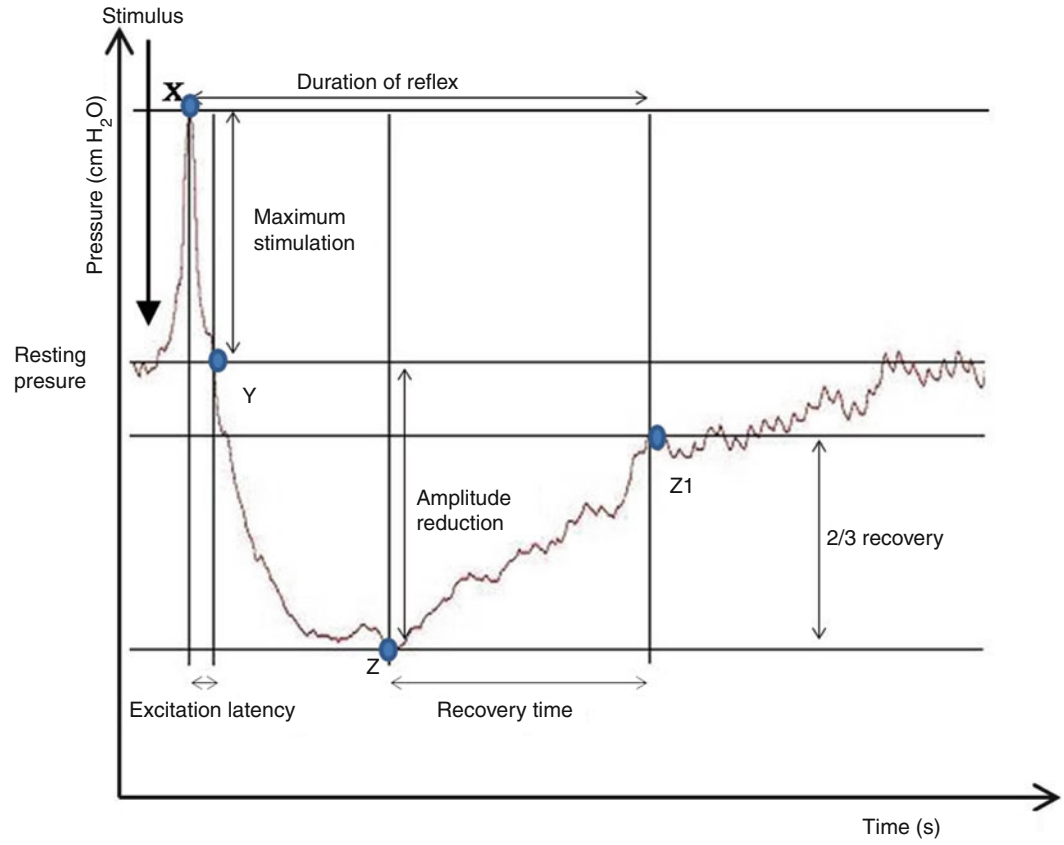

Fig. 11.3 The various components of the RAIR reflex on a manometry trace: (**a**) Excitation peak: initial increase in the resting pressure associated with the sudden rectal distension. (**b**) Latency (*X–Y*): duration from the point of excitation peak back to the baseline pressure. (**c**) Point of maximum relaxation (*Z*): lowest point of resting pressure secondary to reflex IAS relaxation. (**d**) Recovery time (*Z–Z1*): the duration between maximum relaxation and the point at which the resting pressure recovers to two thirds its baseline. (**e**) Total reflex duration (*X–Z1*): calculated as the duration from the point of the Excitation Peak to the point two thirds' the recovery (*Z1*)

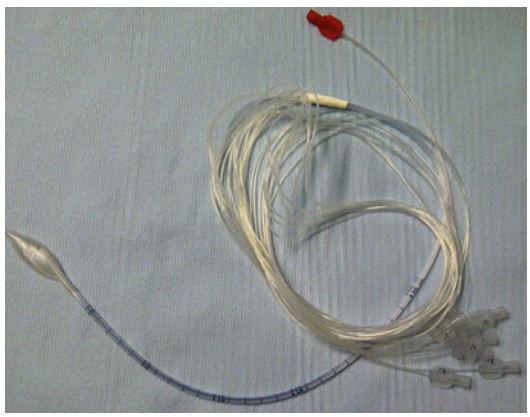

Fig. 11.4 Eight channel manometry catheter with a balloon

disturbed continence. Kaur et al. showed significantly greater sphincter relaxation in the incontinent patients as compared with constipated patients or control subjects [43]. Others have shown that the volume required to induce the reflex was lower in incontinent patients than controls [18, 44].

Anal Sensory Testing

The anal canal mucosa has a plethora of afferent nerve endings allowing the perception of wide range of sensory modalities including touch, pain and temperature [45]. Intact anal sensory function is vital to the continence mechanism.

Technique

Intactness of anal sensory function is clinically tested using electric stimulation. A bipolar electrode and a signal generator are used to provide a wave impulse at 5 Hz of 100 μs duration. The electrode catheter is placed in the anal canal and the current is incrementally increased (up to a maximum of 20 mA) until the subject reports a tingling or pricking sensation. The lowest recorded current of three readings is noted as the sensory threshold [46].

Clinical Utility

Normal electrical sensory threshold in the anal canal is 4 mA (2–7 mA) [46, 47]. Thresholds increase with age and thickness of the subepithelial layer of the anal canal [47]. The primary role of testing for anal sensations in the context of assessment of faecal incontinence is in identifying hindgut (somatic) denervation which can be present in neurogenic incontinence (e.g. in spinal cord injury, multiple sclerosis and diabetic neuropathy) [21, 48–50].

Rectal Sensory Testing

Intact rectal sensory function is an integral component of the mechanism of continence. In contrast to the anal canal, the modalities of rectal sensations are indistinct (visceral sensations) and the rectum is insensitive to stimuli capable of producing pain and other sensations when applied to a somatic cutaneous surface [45, 51]. Full understanding of the types and functions of rectal receptors is still lacking [45, 52]. Rectal distension is associated with the perception of rectal filling and the coordination of specific anorectal reflexes, and although the nature of the mediating receptors is not fully delineated, specialised mechanoreceptors are most probably responsible [52]. Intraganglionic Laminar Endings (IGLEs) are known to be present in the upper gastrointestinal tract; however, recent animal work has revealed that the rectum contains morphologically and functionally unique IGLEs [53]. In clinical practice, testing rectal sensations is performed by two ways (mucosal electrosensitivity and balloon distension). A recent study showed good reproducibility of a newly designed multimodal rectal probe used to test electrical, thermal and mechanical sensory modalities in the rectum [54].

Electric Sensory Thresholds

Technique

This is assessed using a bipolar electrode catheter and pulse generator similar to those used for anal sensory testing. However, the stimulus used is modified to the frequency of 10 Hz and the duration of 500 μs. The threshold is determined by gradually increasing the stimulus (up to a maximum of 50 mA) until the subject reports perception of lower abdominal discomfort or colicky sensation [46, 55].

Clinical Utility

Normal electric sensory thresholds in the rectum range widely between 2.9 and 53.0 mA and is usually higher in females in comparison with males [56]. Patients with neurogenic incontinence have impaired sensation to electrical stimulation [49]. In a study of 68 subjects, Meagher et al. demonstrated circumferential variation of rectal electric sensory threshold suggesting that it is the pelvic floor rather than rectal mucosa which is stimulated with this testing modality [57].

Balloon Distension Thresholds

Technique

Rectal distension thresholds can be tested either by manually distending a latex rectal balloon or by distending a polyethylene rectal balloon using a barostat device [30, 55]. The use of the barostat permits more controlled distensions and therefore provides more physiological and less rater-dependent assessment. Moreover, it enables the measurement of changes in both pressure and volume on distension, therefore permitting assessment of pressure-volume relationships and rectal compliance [58].

The distension thresholds assessed clinically include [7, 9, 59–61] (a) first sensation (FS) (first awareness of the intrarectal balloon), (b) desire to defecate (DD), (c) urgency to defecate (U) and (d) maximally tolerated volume (MTV) (the volume which can be tolerated in total before pain occurs).

Clinical Utility

Sensory thresholds to rectal distension in asymptomatic subjects vary significantly between different centres [7, 9, 59–61]. The pattern of balloon distension [62] as well as age of the subject [9] affects rectal perception. The large inter- and intra-subject variation in values and the wide normal range impact the clinical value of these measurements [63]. Moreover, thresholds for rectal distension may be elevated, normal or reduced in faecal incontinence [16, 17]. Nevertheless, evidence of loss or significant reduction of rectal perception to balloon distension (hindgut denervation) and conversely the evidence of exaggerated rectal sensation (rectal hypersensitivity and/or reduced rectal capacity) are important findings when assessing faecal incontinence. Impaired rectal sensation can be associated with symptoms of incontinence [64, 65], and there is evidence that sensory retraining can improve symptoms [65, 66]. On another hand, some patients with urge faecal incontinence have significantly reduced sensory thresholds to rectal distension; this rectal hypersensitivity has been found to be associated with reduced rectal compliance and exaggerated rectosigmoid motor activity highlighting a specific entity of incontinent patients [17, 67].

Rectal Compliance Measurement

Given a slow filling rate, the intraluminal rectal pressure does not increase until the maximum tolerated volume is approached; this phenomenon of receptive relaxation (compliance) of the rectal wall defines the rectum as a reservoir.

Technique

Rectal compliance is measured by assessing rectal pressure-volume relationships, either by manually inflating a latex balloon inside the rectum with air or water or by mechanically distending an infinitely compliant bag using the barostat device. The latter is the preferable technique as it utilises a controlled rate of distension and an infinitely compliant bag, eliminating technical sources of variability [6, 58, 68]. The polyethylene barostat bag has to be infinitely compliant, either a bag infinitely compliant up to its maximum volume or a fixed large-volume noncompliant bag which has a maximum volume greater than the maximum volume of the rectum [69]. Mechanical instillation of air into the bag by the barostat device allows for the standardisation of flow rate and accurate measurements of volume-pressure relationship. Specific sequence of distension protocols is used and the pressure-volume data obtained is plotted on special software to calculate compliance (change in volume divided by change in pressure).

Clinical Utility

Rectal compliance is a measure of the combined sensorimotor function of the rectum. Barostat measurement of rectal compliance using identical distension protocols has been shown to have good inter- and intra-subject reproducibility [6, 58, 68]. Abnormal compliance has been demonstrated in patients with various anorectal dysfunction, but interpretation of results can be problematic due to the lack of standardised protocols for measurement and the contribution of abnormal rectal sensations [52]. Normal rectal compliance is 11.9 ± 4.1 mL/mmHg [58]. Rectal compliance is reduced in patients with colitis [70] and in some patients with systemic sclerosis [71, 72].

Symptoms of faecal urgency and urge incontinence may be associated with reduced compliance and rectal capacity [17, 67, 73], however, changes in compliance may be secondary to the incontinence and not causative [73]. Moreover, the contributory factor of changes in the rectal sensory function makes interpretation difficult. Some studies have suggested that reduced compliance is associated with reduced rectal sensitivity [74, 75]. Nevertheless, other studies in healthy subjects [76], patients following radiotherapy [77] and ulcerative proctitis [70] suggest that reduced rectal compliance is associated with and perhaps contributes to rectal hypersensitivity.

Electrophysiology

Neurophysiological tests of the anorectum include assessment of the conduction of the pudendal nerve and electromyography of the

external sphincter. These tests are primarily employed in the research setting and their clinical utility is limited.

Pudendal Nerve Terminal Motor Latency (PNTML)

This is intended to assess conduction in the pudendal nerve which supplies the EAS. The technique involves the use of an integrated stimulation and recording electrode mounted on a gloved finger and placed directly over the nerve as it passes over the ischial spine in the lateral pelvic wall [78]. In theory, prolonged latency between stimulating the nerve and the beginning of EAS contraction indicates pudendal nerve damage or neuropathy. However, the use of this test to indicate pudendal neuropathy has been challenged as, firstly, the technique is highly operator-dependent and its reproducibility is unknown [30]; secondly, the measured latency is a reflection of the fastest conducting fibres and hence latencies may be normal despite the presence of significant neuropathy. Moreover, there is poor correlation between prolonged PNTML and anal sphincter pressures [79]. PNTML cannot be recommended for the routine evaluation of patients with faecal incontinence [80].

Electromyography (EMG) of the External Anal Sphincter

Electromyography of the EAS and puborectalis can be performed to identify areas of sphincter injury by mapping the sphincter (concentric needle EMG) or to identify the characteristic denervation-reinnervation patterns indicative of nerve injury (small needle EMG: single-fibre studies); however, the technique is invasive and a potential source of introducing infection. For the purpose of identifying EAS trauma, anal endosonography has largely replaced needle EMG as a much better tolerated and more sensitive test [81, 82]. However, the use of non-invasive (surface or intra-anal) EMG electrodes to identify sphincter contraction remains a useful component of biofeedback treatment [83–85].

Other Functional Tests

Rectal Balloon Expulsion Test

This is a useful screening test for evacuation disorders and is not performed routinely in the course of assessment of faecal incontinence. The ability of the patient to expel a water-filled intrarectal balloon whilst seated on a commode in a private setting is assessed. The balloon is inflated either by a fixed volume [5] or alternatively with volume which elicits the desire to defecate [86].

Stool-Substitute Retention Test

This test has been suggested as a tool to evaluate anorectal function in patients prior to the consideration of reversal of a defunctioning stoma [87]. It involves the instillation of a specific volume of material simulating semi-formed consistency stool (e.g. porridge) into the rectum to assess the volumes tolerated, the extent of any leakage on lying, standing and walking and the ability to hold the contents for 30 min [87]. Saline has been used in this way to assess anorectal function (saline infusion test) [18, 88–90]; however, it assesses the capacity to maintain continence during conditions that simulate diarrhoea.

Imaging Studies

Imaging of the sphincter and the pelvic floor combined with anorectal physiology provides an integrated assessment of sphincter function and aids treatment planning. In this section, a brief summary of the most widely used imaging modalities is given. These provide either static anatomical information or more dynamic or functional information.

Static Imaging (Tests of Structure)

Endoanal Ultrasound

Endoanal ultrasound assessment is the most reliable and least invasive modality for defining structural sphincter defects [80, 91, 92].

Technique

Although transperineal and transvaginal approaches to ultrasound sphincter imaging are feasible, endoprobes have the advantage of imaging in the ideal anatomical plane and are the gold standard [92, 93]. In males, examination in the left lateral position is adequate, but in females, either the prone or lithotomy position is preferred, as in the left lateral position the anus becomes deformed as the pelvic organs pull it over with subsequent loss of the symmetry of the anterior structures [94]. A rotating 7 MHz or high frequency (10–15 MHz) transducer providing a 360º view is generally used [95, 96]. Image analysis is usually performed using sequential axial images through the anal canal, although a 3D data volume acquisition is available in certain machines allowing image-analysis in the coronal and sagittal planes [97].

Clinical Utility

Endoanal ultrasound assesses the thickness of the sphincter muscles, the presence of atrophy as well as the integrity of the sphincters and the presence of muscle tearing or injury. The axial image is composite of reflections from individual anatomical layers and interfaces between layers. On ultrasound, the anus is fundamentally comprised of four layers, from inside outwards: subepithelium, internal anal sphincter, longitudinal muscle layer and the external anal sphincter [93] (Fig. 11.5).

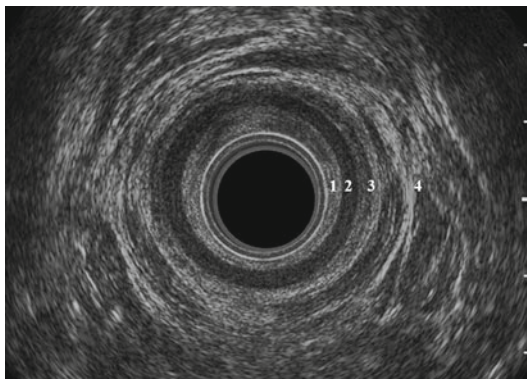

Fig. 11.5 Endoanal ultrasound image showing the various layers seen: *1* Subepithelium, *2* IAS, *3* Longitudinal muscle layer, *4* EAS

The normal internal anal sphincter (IAS) varies in thickness with age, measuring 1–2 mm in young adults and up to 4 mm in the elderly [93, 98]. Abnormally increased internal sphincter thickness is associated with high-grade rectal intussusception and rectal prolapse [99, 100] and is an indication for proctography to exclude rectal prolapse if the diagnosis has not been made clinically [93]. On the contrary, an abnormally thin internal sphincter denotes atrophy and is often seen in patients with passive faecal incontinence [101]. Endoanal ultrasound also assesses the integrity of the internal sphincter, and evidence of disruption can be found in patients who had previously undergone lateral sphincterotomy [102, 103] or stretch procedures [104, 105]. Moreover, internal sphincter disruption can be related to inadvertent injury during haemorrhoidectomy [106]. Endosonographic evidence of IAS injury is predictive of faecal incontinence following obstetric anal sphincter injury [107].

Abnormalities involving the external anal sphincter (EAS) are similarly those of tearing or abnormal muscle density. Tearing of the external sphincter is most commonly related to childbirth, and endosonography demonstrates the ruptured area as a relatively homogenous low-reflectivity segment within the external sphincter, usually involving other layers, particularly the longitudinal layer and often the internal sphincter [93]. External sphincter atrophy is difficult to recognise on endosonography as it involves loss of muscle fibres with fat replacement to the extent that the acoustic interface on the outer border of the sphincter is lost making a precise measurement of the thickness of the sphincter impossible.

Magnetic Resonance Imaging (MRI)

High-resolution MRI techniques have enabled the detailed examination and delineation of injury to the muscles of the pelvic floor. The study is performed using a surface coil placed on the pelvis of the patient; however, an endocoil can be used to obtain detailed images of the sphincter complex (endoanal MRI).

MRI of the pelvic floor is indicated in patients with pelvic floor dysfunction to identify morphologic

defects in the levator ani muscles with the rationale that patients might present with symptoms isolated to one pelvic compartment but may have concomitant defects in other compartments [108]. Defective levator ani muscle is highly associated with pelvic floor dysfunction [109, 110] and specifically pelvic organ prolapse [111]; therefore, pelvic floor MRI in the context of faecal incontinence assessment is important in the subgroup of patients who are suspected to have those clinical conditions. Moreover, there is evidence that surgery for pelvic organ prolapse has higher failure rates in patients with levator ani impairment [112, 113]. With regard to the sole purpose of assessing the sphincter, numerous studies have found comparable performance of endosonography and MRI for sphincter tears [114–117], with a preference for endosonography for the internal sphincter but a definite advantage for MRI in detecting external sphincter atrophy.

Dynamic Imaging (Tests of Structure and Function)

Dynamic imaging of the pelvic floor involves defecography (proctography), and it allows the global assessment of the pelvic floor to detect abnormalities of structural descent, prolapse or evacuation difficulties. It provides functional information on the pelvic floor during a provocatory manoeuvre such as straining, rectal evacuation or pelvic floor contraction.

Proctography has significantly evolved since it was first described by Burhenne in 1964 [118]. Traditionally, it was limited to the fluoroscopic technique; however, with the recent improvement in MRI technology and the development of rapid image acquisition techniques, dynamic MR proctography is increasingly replacing the conventional techniques.

As pelvic floor weakness and dysfunction usually involves more than one compartment, imaging of the compartments is best done simultaneously. To achieve this, conventionally, evacuation proctography is modified by the additional opacification of the bladder and small bowel [119–121]. However, the development of MR proctography allows pelvic floor motion to be visualised together with the definition of the anatomy of all pelvic organs and muscular structures in a superior way which does not involve ionising radiation. The main purposes of proctography are to provide images of rectal configuration throughout the phases of evacuation and also to provide an assessment of whether voiding is normal or prolonged [122]. The assessment of completeness of evacuation and its rate is considered an essential part of the examination [123]. Although, proctography is most useful for demonstrating pelvic organ prolapse in suspected patients [124, 125]; recent studies show that performing the investigation in patients presenting with faecal incontinence only reveal significant pelvic floor abnormalities which might be contributing to multifactorial incontinence [126, 127] and have bearing on the surgical decision for management [126].

Conclusion

There is a variety of anorectal tests which can be used in the context of assessment of faecal incontinence. Although they do not provide quantification or good correlation with the clinical status of the patient, they give a useful insight into the potential underlying pathophysiology. Multiple tests examining various aspects of function and structure of the anorectum are usually required to complete the assessment, as the pathophysiology in faecal incontinence is often multifactorial. Revealing the pattern of those multiple sensorimotor dysfunctions in patients with faecal incontinence confirms the underlying disorder and guides treatment options (Fig. 11.6).

On the other hand, clinical assessment remains instrumental in the assessment of the condition and its possible aetiology. Firstly, the clinician should ensure the absence of any suspicion of mucosal pathology and take the appropriate investigative steps if this is present. Clinical assessment provides information on the severity of symptoms and the extent of disruption to the quality of life which is important in planning treatment measures.

Basic physiological tests should be routine in all cases of faecal incontinence. Although normal data is relatively lacking [7, 8] and values overlap between normal and incontinent

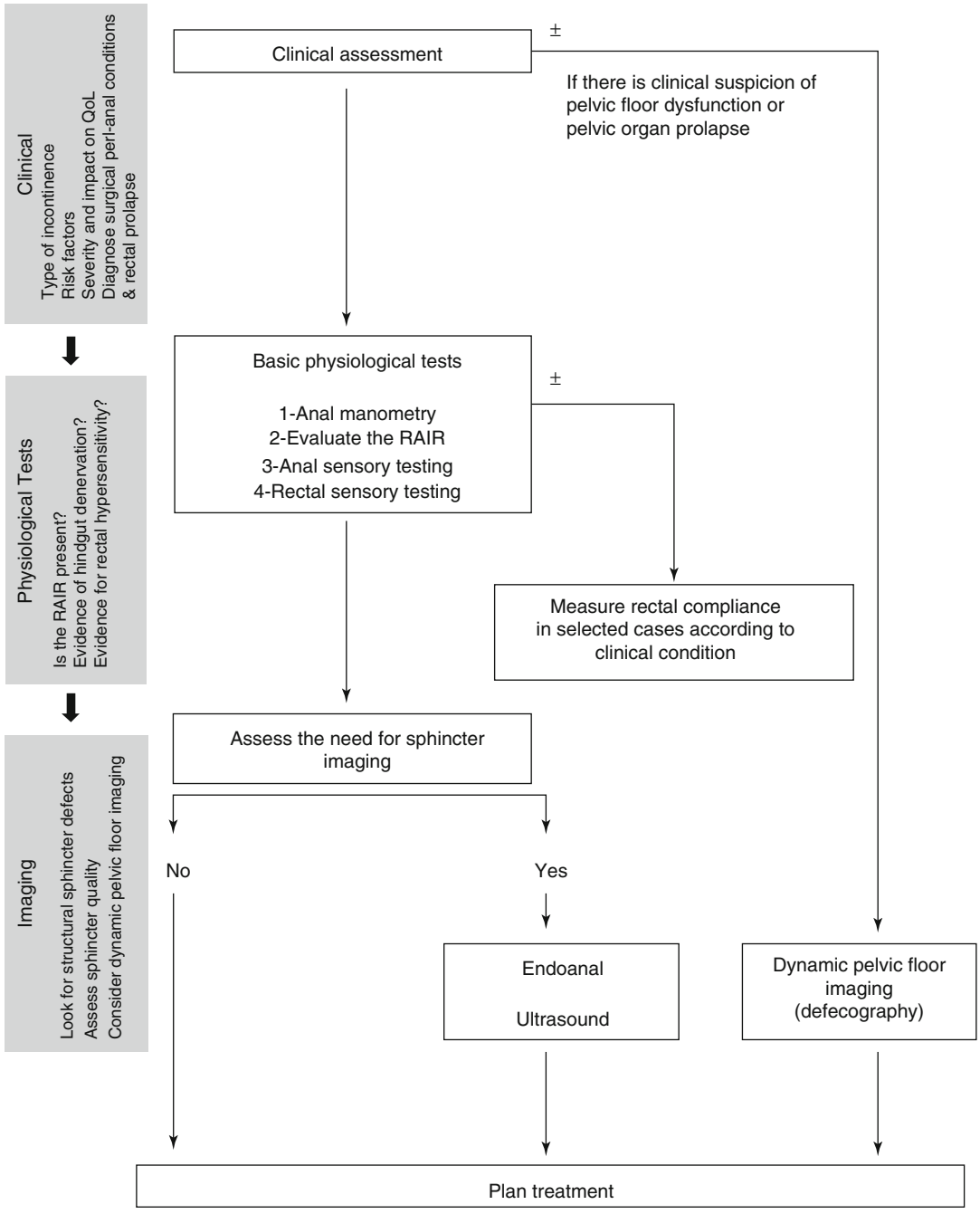

Fig. 11.6 An algorithm highlighting the approach to assessment of faecal incontinence

subjects [19], physiological tests for the individual patient can be very useful in highlighting the underlying dysfunction. Routine physiological tests in incontinent patients should include anal manometry, RAIR evaluation and the testing of anal and rectal sensory thresholds. Rectal compliance measurement is probably required only in selected cases (Table 11.3).

The need for imaging of the sphincter is dictated by the clinical condition and the suspicion

Table 11.3 Summary of basic anorectal physiology tests

Test	Details	Relevance in faecal incontinence
Anal manometry	Measurement of	Usually pressures are reduced
	Resting pressure	Reduced resting pressure → IAS dysfunction
	Squeeze pressure	Reduced squeeze pressure → EAS dysfunction
	Cough reflex	
RAIR	Sudden rectal distension to elicit reflex IAS relaxation	Absent in Hirschsprung's disease
Anal sensory testing	Electric sensory thresholds	Somatic denervation denotes neurogenic element
		Impaired anal sensations means certain interventions can be feasible (e.g. anal plugs)
Rectal sensory testing	1. Electric sensory thresholds	Higher distension thresholds suggest impaired rectal sensation
	2. Balloon distension thresholds	Hindgut denervation in neurogenic incontinence
		Rectal hypersensitivity can be associated with urge incontinence
		Informs biofeedback strategies
Rectal compliance	Measure change in volume over change in pressure during rectal distension	Reduced compliance can be associated with rectal hypersensitivity
		Changes in compliance may point to certain aetiologies (e.g. reduced compliance in patients with colitis, radiation proctitis and connective tissue disorders)

of a sphincter injury; however, it is advisable to routinely perform endoanal ultrasound as this enables the identification of a potentially repairable sphincter defect. In elderly patients, those who have symptoms of difficulty in evacuation or in situations where pelvic organ prolapse is clinically suspected, it is prudent to proceed with dynamic pelvic floor imaging to enable effective and accurate management planning.

References

1. Rao SS. Diagnosis and management of fecal incontinence. American College of Gastroenterology Practice Parameters Committee. Am J Gastroenterol. 2004;99(8):1585–604.
2. Jorge JM, Wexner SD. Etiology and management of fecal incontinence. Dis Colon Rectum. 1993;36(1):77–97.
3. Pescatori M, Anastasio G, Bottini C, Mentasti A. New grading and scoring for anal incontinence. Evaluation of 335 patients. Dis Colon Rectum. 1992;35(5):482–7.
4. Vaizey CJ, Carapeti E, Cahill JA, Kamm MA. Prospective comparison of faecal incontinence grading systems. Gut. 1999;44(1):77–80.
5. Rao SS, Azpiroz F, Diamant N, Enck P, Tougas G, Wald A. Minimum standards of anorectal manometry. Neurogastroenterol Motil. 2002;14(5):553–9.
6. Bharucha AE, Seide B, Fox JC, Zinsmeister AR. Day-to-day reproducibility of anorectal sensorimotor assessments in healthy subjects. Neurogastroenterol Motil. 2004;16(2):241–50.
7. Rao SS, Hatfield R, Soffer E, Rao S, Beaty J, Conklin JL. Manometric tests of anorectal function in healthy adults. Am J Gastroenterol. 1999;94(3):773–83.
8. Diamant NE, Kamm MA, Wald A, Whitehead WE. AGA technical review on anorectal testing techniques. Gastroenterology. 1999;116(3):735–60.
9. Bannister JJ, Abouzekry L, Read NW. Effect of aging on anorectal function. Gut. 1987;28(3):353–7.
10. Enck P, Kuhlbusch R, Lubke H, Frieling T, Erckenbrecht JF. Age and sex and anorectal manometry in incontinence. Dis Colon Rectum. 1989;32(12):1026–30.
11. Loening-Baucke V, Anuras S. Effects of age and sex on anorectal manometry. Am J Gastroenterol. 1985;80(1):50–3.
12. Laurberg S, Swash M, Henry MM. Delayed external sphincter repair for obstetric tear. Br J Surg. 1988;75(8):786–8.
13. Taylor BM, Beart Jr RW, Phillips SF. Longitudinal and radial variations of pressure in the human anal sphincter. Gastroenterology. 1984;86(4):693–7.
14. Sun WM, Read NW. Anorectal function in normal human subjects: effect of gender. Int J Colorectal Dis. 1989;4(3):188–96.
15. Jorge JM, Wexner SD. Anorectal manometry: techniques and clinical applications. South Med J. 1993;86(8):924–31.
16. Sun WM, Donnelly TC, Read NW. Utility of a combined test of anorectal manometry, electromyography,

and sensation in determining the mechanism of 'idiopathic' faecal incontinence. Gut. 1992;33(6):807–13.
17. Bharucha AE, Fletcher JG, Harper CM, Hough D, Daube JR, Stevens C, et al. Relationship between symptoms and disordered continence mechanisms in women with idiopathic faecal incontinence. Gut. 2005;54(4):546–55.
18. Read NW, Harford WV, Schmulen AC, Read MG, Santa Ana C, Fordtran JS. A clinical study of patients with fecal incontinence and diarrhea. Gastroenterology. 1979;76(4):747–56.
19. McHugh SM, Diamant NE. Effect of age, gender, and parity on anal canal pressures. Contribution of impaired anal sphincter function to fecal incontinence. Dig Dis Sci. 1987;32(7):726–36.
20. Delechenaut P, Leroi AM, Weber J, Touchais JY, Czernichow P, Denis P. Relationship between clinical symptoms of anal incontinence and the results of anorectal manometry. Dis Colon Rectum. 1992; 35(9):847–9.
21. MacDonagh R, Sun WM, Thomas DG, Smallwood R, Read NW. Anorectal function in patients with complete supraconal spinal cord lesions. Gut. 1992; 33(11):1532–8.
22. Sun WM, MacDonagh R, Forster D, Thomas DG, Smallwood R, Read NW. Anorectal function in patients with complete spinal transection before and after sacral posterior rhizotomy. Gastroenterology. 1995;108(4):990–8.
23. Bharucha AE. Outcome measures for fecal incontinence: anorectal structure and function. Gastroenterology. 2004;126(1 Suppl 1):S90–8.
24. Carrington EV, Knowles CH. The influence of sacral nerve stimulation on anorectal dysfunction. Colorectal Dis. 2011;13 Suppl 2:5–9.
25. Roberts JP, Williams NS. The role and technique of ambulatory anal manometry. Baillieres Clin Gastroenterol. 1992;6(1):163–78.
26. Kumar D, Waldron D, Williams NS, Browning C, Hutton MR, Wingate DL. Prolonged anorectal manometry and external anal sphincter electromyography in ambulant human subjects. Dig Dis Sci. 1990;35(5):641–8.
27. Braun JC, Treutner KH, Dreuw B, Klimaszewski M, Schumpelick V. Vectormanometry for differential diagnosis of fecal incontinence. Dis Colon Rectum. 1994;37(10):989–96.
28. Perry RE, Blatchford GJ, Christensen MA, Thorson AG, Attwood SE. Manometric diagnosis of anal sphincter injuries. Am J Surg. 1990;159(1):112–6; discussion 6–7.
29. Yang YK, Wexner SD. Anal pressure vectography is of no apparent benefit for sphincter evaluation. Int J Colorectal Dis. 1994;9(2):92–5.
30. Bharucha AE. Update of tests of colon and rectal structure and function. J Clin Gastroenterol. 2006; 40(2):96–103.
31. Schizas AM, Emmanuel AV, Williams AB. Anal canal vector volume manometry. Dis Colon Rectum. 2011;54(6):759–68.
32. Jones MP, Post J, Crowell MD. High-resolution manometry in the evaluation of anorectal disorders: a simultaneous comparison with water-perfused manometry. Am J Gastroenterol. 2007;102(4):850–5.
33. Miller R, Bartolo DC, Cervero F, Mortensen NJ. Anorectal sampling: a comparison of normal and incontinent patients. Br J Surg. 1988;75(1):44–7.
34. Lubowski DZ, Nicholls RJ, Swash M, Jordan MJ. Neural control of internal anal sphincter function. Br J Surg. 1987;74(8):668–70.
35. Goes RN, Simons AJ, Masri L, Beart Jr RW. Gradient of pressure and time between proximal anal canal and high-pressure zone during internal anal sphincter relaxation. Its role in the fecal continence mechanism. Dis Colon Rectum. 1995;38(10):1043–6.
36. Gowers WR. The automatic action of the sphincter ani. Proc Royal Soc London. 1877;26:77–84.
37. Denny-Brown D, Robertson EG. An investigation of the nervous control of defaecation. Brain. 1935;58(2): 256–310.
38. Lawson JO, Nixon HH. Anal canal pressures in the diagnosis of Hirschsprung's disease. J Pediatr Surg. 1967;2(6):544–52.
39. Howard ER, Nixon HH. Internal anal sphincter. Observations on development and mechanism of inhibitory responses in premature infants and children with Hirschprung's disease. Arch Dis Child. 1968; 43(231):569–78.
40. Meunier P, Marechal JM, Mollard P. Accuracy of the manometric diagnosis of Hirschsprung's disease. J Pediatr Surg. 1978;13(4):411–5.
41. O'Riordain MG, Molloy RG, Gillen P, Horgan A, Kirwan WO. Rectoanal inhibitory reflex following low stapled anterior resection of the rectum. Dis Colon Rectum. 1992;35(9):874–8.
42. Lewis WG, Williamson ME, Miller AS, Sagar PM, Holdsworth PJ, Johnston D. Preservation of complete anal sphincteric proprioception in restorative proctocolectomy: the inhibitory reflex and fine control of continence need not be impaired. Gut. 1995;36(6):902–6.
43. Kaur G, Gardiner A, Duthie GS. Rectoanal reflex parameters in incontinence and constipation. Dis Colon Rectum. 2002;45(7):928–33.
44. Sun WM, Read NW, Donnelly TC. Anorectal function in incontinent patients with cerebrospinal disease. Gastroenterology. 1990;99(5):1372–9.
45. Duthie HL, Gairns FW. Sensory nerve-endings and sensation in the anal region of man. Br J Surg. 1960;47:585–95.
46. Roe AM, Bartolo DC, Mortensen NJ. New method for assessment of anal sensation in various anorectal disorders. Br J Surg. 1986;73(4):310–2.
47. Felt-Bersma RJ, Poen AC, Cuesta MA, Meuwissen SG. Anal sensitivity test: what does it measure and do we need it? Cause or derivative of anorectal complaints. Dis Colon Rectum. 1997;40(7):811–6.
48. Rogers J, Levy DM, Henry MM, Misiewicz JJ. Pelvic floor neuropathy: a comparative study of diabetes mellitus and idiopathic faecal incontinence. Gut. 1988;29(6):756–61.

49. Speakman CT, Kamm MA. Abnormal visceral autonomic innervation in neurogenic faecal incontinence. Gut. 1993;34(2):215–21.
50. Greving I, Tegenthoff M, Nedjat S, Orth G, Botel U, Meister V, et al. Anorectal functions in patients with spinal cord injury. Neurogastroenterol Motil. 1998;10(6):509–15.
51. Rogers J. Testing for and the role of anal and rectal sensation. Baillieres Clin Gastroenterol. 1992;6(1):179–91.
52. Bajwa A, Emmanuel A. The physiology of continence and evacuation. Best Pract Res Clin Gastroenterol. 2009;23(4):477–85.
53. Lynn PA, Olsson C, Zagorodnyuk V, Costa M, Brookes SJ. Rectal intraganglionic laminar endings are transduction sites of extrinsic mechanoreceptors in the guinea pig rectum. Gastroenterology. 2003; 125(3):786–94.
54. Brock C, Nissen TD, Gravesen FH, Frokjaer JB, Omar H, Gale J, et al. Multimodal sensory testing of the rectum and rectosigmoid: development and reproducibility of a new method. Neurogastroenterol Motil. 2008;20(8):908–18.
55. Kamm MA, Lennard-Jones JE. Rectal mucosal electrosensory testing – evidence for a rectal sensory neuropathy in idiopathic constipation. Dis Colon Rectum. 1990;33(5):419–23.
56. Jameson JS, Chia YW, Kamm MA, Speakman CT, Chye YH, Henry MM. Effect of age, sex and parity on anorectal function. Br J Surg. 1994;81(11):1689–92.
57. Meagher AP, Kennedy ML, Lubowski DZ. Rectal mucosal electrosensitivity – what is being tested? Int J Colorectal Dis. 1996;11(1):29–33.
58. Fox M, Thumshirn M, Fried M, Schwizer W. Barostat measurement of rectal compliance and capacity. Dis Colon Rectum. 2006;49(3):360–70.
59. Ryhammer AM, Laurberg S, Bek KM. Age and anorectal sensibility in normal women. Scand J Gastroenterol. 1997;32(3):278–84.
60. Sloots CE, Felt-Bersma RJ, Cuesta MA, Meuwissen SG. Rectal visceral sensitivity in healthy volunteers: influences of gender, age and methods. Neurogastroenterol Motil. 2000;12(4):361–8.
61. Bharucha AE, Fletcher JG, Seide B, Riederer SJ, Zinsmeister AR. Phenotypic variation in functional disorders of defecation. Gastroenterology. 2005; 128(5):1199–210.
62. Sun WM, Read NW, Prior A, Daly JA, Cheah SK, Grundy D. Sensory and motor responses to rectal distention vary according to rate and pattern of balloon inflation. Gastroenterology. 1990;99(4):1008–15.
63. Kendall GP, Thompson DG, Day SJ, Lennard-Jones JE. Inter- and intraindividual variation in pressure-volume relations of the rectum in normal subjects and patients with the irritable bowel syndrome. Gut. 1990;31(9):1062–8.
64. Sun WM, Read NW, Miner PB. Relation between rectal sensation and anal function in normal subjects and patients with faecal incontinence. Gut. 1990;31(9): 1056–61.
65. Buser WD, Miner Jr PB. Delayed rectal sensation with fecal incontinence. Successful treatment using anorectal manometry Gastroenterology. 1986;91(5):1186–91.
66. Wald A, Tunuguntla AK. Anorectal sensorimotor dysfunction in fecal incontinence and diabetes mellitus. Modification with biofeedback therapy. N Engl J Med. 1984;310(20):1282–7.
67. Chan CL, Lunniss PJ, Wang D, Williams NS, Scott SM. Rectal sensorimotor dysfunction in patients with urge faecal incontinence: evidence from prolonged manometric studies. Gut. 2005;54(9):1263–72.
68. Cremonini F, Houghton LA, Camilleri M, Ferber I, Fell C, Cox V, et al. Barostat testing of rectal sensation and compliance in humans: comparison of results across two centres and overall reproducibility. Neurogastroenterol Motil. 2005;17(6):810–20.
69. Krogh K, Ryhammer AM, Lundby L, Gregersen H, Laurberg TS. Comparison of methods used for measurement of rectal compliance. Dis Colon Rectum. 2001;44(2):199–206.
70. Rao SS, Read NW, Davison PA, Bannister JJ, Holdsworth CD. Anorectal sensitivity and responses to rectal distention in patients with ulcerative colitis. Gastroenterology. 1987;93(6):1270–5.
71. Sallam HS, McNearney TA, Chen JZ. Anorectal motility and sensation abnormalities and its correlation with anorectal symptoms in patients with systemic sclerosis: a preliminary study. ISRN Gastroenterol. 2011;2011:402–583.
72. Forbes A, Marie I. Gastrointestinal complications: the most frequent internal complications of systemic sclerosis. Rheumatology (Oxford). 2009;48 Suppl 3:iii36–9.
73. Rasmussen O, Christensen B, Sorensen M, Tetzschner T, Christiansen J. Rectal compliance in the assessment of patients with fecal incontinence. Dis Colon Rectum. 1990;33(8):650–3.
74. Siproudhis L, Bellissant E, Juguet F, Allain H, Bretagne JF, Gosselin M. Perception of and adaptation to rectal isobaric distension in patients with faecal incontinence. Gut. 1999;44(5):687–92.
75. Salvioli B, Bharucha AE, Rath-Harvey D, Pemberton JH, Phillips SF. Rectal compliance, capacity, and rectoanal sensation in fecal incontinence. Am J Gastroenterol. 2001;96(7):2158–68.
76. Malcolm A, Phillips SF, Camilleri M, Hanson RB. Pharmacological modulation of rectal tone alters perception of distention in humans. Am J Gastroenterol. 1997;92(11):2073–9.
77. Varma JS, Smith AN, Busuttil A. Correlation of clinical and manometric abnormalities of rectal function following chronic radiation injury. Br J Surg. 1985;72(11):875–8.
78. Rogers J, Henry MM, Misiewicz JJ. Disposable pudendal nerve stimulator: evaluation of the standard instrument and new device. Gut. 1988;29(8):1131–3.
79. Wexner SD, Marchetti F, Salanga VD, Corredor C, Jagelman DG. Neurophysiologic assessment of the anal sphincters. Dis Colon Rectum. 1991;34(7):606–12.

80. Barnett JL, Hasler WL, Camilleri M. American Gastroenterological Association medical position statement on anorectal testing techniques. American Gastroenterological Association. Gastroenterology. 1999;116(3):732–60.
81. Enck P, von Giesen HJ, Schafer A, Heyer T, Gantke B, Flesch S, et al. Comparison of anal sonography with conventional needle electromyography in the evaluation of anal sphincter defects. Am J Gastroenterol. 1996;91(12):2539–43.
82. Law PJ, Kamm MA, Bartram CI. A comparison between electromyography and anal endosonography in mapping external anal sphincter defects. Dis Colon Rectum. 1990;33(5):370–3.
83. Patankar SK, Ferrara A, Larach SW, Williamson PR, Perozo SE, Levy JR, et al. Electromyographic assessment of biofeedback training for fecal incontinence and chronic constipation. Dis Colon Rectum. 1997;40(8):907–11.
84. Mahony RT, Malone PA, Nalty J, Behan M, O'Connell PR, O'Herlihy C. Randomized clinical trial of intra-anal electromyographic biofeedback physiotherapy with intra-anal electromyographic biofeedback augmented with electrical stimulation of the anal sphincter in the early treatment of postpartum fecal incontinence. Am J Obstet Gynecol. 2004;191(3):885–90.
85. Voorham-van der Zalm PJ, Voorham JC, van den Bos TW, Ouwerkerk TJ, Putter H, Wasser MN, et al. Reliability and differentiation of pelvic floor muscle electromyography measurements in healthy volunteers using a new device: the multiple array probe leiden (MAPLe). Neurourol Urodyn. 2013;32(4):341–8.
86. Minguez M, Herreros B, Sanchiz V, Hernandez V, Almela P, Anon R, et al. Predictive value of the balloon expulsion test for excluding the diagnosis of pelvic floor dyssynergia in constipation. Gastroenterology. 2004;126(1):57–62.
87. Pares D, Duncan J, Dudding T, Phillips RK, Norton C. Investigation to predict faecal continence in patients undergoing reversal of a defunctioning stoma (porridge enema test). Colorectal Dis. 2008;10(4):379–85.
88. Rao SS, Read NW, Stobart JA, Haynes WG, Benjamin S, Holdsworth CD. Anorectal contractility under basal conditions and during rectal infusion of saline in ulcerative colitis. Gut. 1988;29(6):769–77.
89. Felt-Bersma RJ, Klinkenberg-Knol EC, Meuwissen SG. Anorectal function investigations in incontinent and continent patients. Differences and discriminatory value. Dis Colon Rectum. 1990;33(6):479–85; discussion 85–6.
90. Kayaba H, Hebiguchi T, Itoh Y, Yoshino H, Mizuno M, Morii M, et al. Evaluation of anorectal function in patients with tethered cord syndrome: saline enema test and fecoflowmetry. J Neurosurg. 2003;98(3 Suppl): 251–7.
91. Rao SS. Pathophysiology of adult fecal incontinence. Gastroenterology. 2004;126(1 Suppl 1):S14–22.
92. Taylor SA. Imaging pelvic floor dysfunction. Best Pract Res Clin Gastroenterol. 2009;23(4):487–503.
93. Bartram C. Endoanal ultrasound. In: Stoker J, Taylor SA, Delancey JOL, editors. Imaging pelvic floor disorders. Berlin: Springer; 2008.
94. Frudinger A, Bartram CI, Halligan S, Kamm M. Examination techniques for endosonography of the anal canal. Abdom Imaging. 1998;23(3):301–3.
95. Law PJ, Kamm MA, Bartram CI. Anal endosonography in the investigation of faecal incontinence. Br J Surg. 1991;78(3):312–4.
96. Bartram C. Radiologic evaluation of anorectal disorders. Gastroenterol Clin North Am. 2001;30(1): 55–75.
97. Gold DM, Bartram CI, Halligan S, Humphries KN, Kamm MA, Kmiot WA. Three-dimensional endoanal sonography in assessing anal canal injury. Br J Surg. 1999;86(3):365–70.
98. Frudinger A, Halligan S, Bartram CI, Price AB, Kamm MA, Winter R. Female anal sphincter: age-related differences in asymptomatic volunteers with high-frequency endoanal US. Radiology. 2002;224(2): 417–23.
99. Dvorkin LS, Chan CL, Knowles CH, Williams NS, Lunniss PJ, Scott SM. Anal sphincter morphology in patients with full-thickness rectal prolapse. Dis Colon Rectum. 2004;47(2):198–203.
100. Marshall M, Halligan S, Fotheringham T, Bartram C, Nicholls RJ. Predictive value of internal anal sphincter thickness for diagnosis of rectal intussusception in patients with solitary rectal ulcer syndrome. Br J Surg. 2002;89(10):1281–5.
101. Vaizey CJ, Kamm MA, Bartram CI. Primary degeneration of the internal anal sphincter as a cause of passive faecal incontinence. Lancet. 1997;349(9052): 612–5.
102. Sultan AH, Kamm MA, Nicholls RJ, Bartram CI. Prospective study of the extent of internal anal sphincter division during lateral sphincterotomy. Dis Colon Rectum. 1994;37(10):1031–3.
103. Garcia-Aguilar J, Belmonte Montes C, Perez JJ, Jensen L, Madoff RD, Wong WD. Incontinence after lateral internal sphincterotomy: anatomic and functional evaluation. Dis Colon Rectum. 1998;41(4): 423–7.
104. Speakman CT, Burnett SJ, Kamm MA, Bartram CI. Sphincter injury after anal dilatation demonstrated by anal endosonography. Br J Surg. 1991;78(12):1429–30.
105. Nielsen MB, Rasmussen OO, Pedersen JF, Christiansen J. Risk of sphincter damage and anal incontinence after anal dilatation for fissure-in-ano. An endosonographic study. Dis Colon Rectum. 1993;36(7):677–80.
106. Abbasakoor F, Nelson M, Beynon J, Patel B, Carr ND. Anal endosonography in patients with anorectal symptoms after haemorrhoidectomy. Br J Surg. 1998;85(11):1522–4.
107. Mahony R, Behan M, Daly L, Kirwan C, O'Herlihy C, O'Connell PR. Internal anal sphincter defect influences continence outcome following obstetric anal sphincter injury. Am J Obstet Gynecol. 2007; 196(3):217.e1–5.

108. Maglinte DD, Kelvin FM, Fitzgerald K, Hale DS, Benson JT. Association of compartment defects in pelvic floor dysfunction. AJR Am J Roentgenol. 1999;172(2):439–44.
109. Morgan DM, Umek W, Stein T, Hsu Y, Guire K, DeLancey JO. Interrater reliability of assessing levator ani muscle defects with magnetic resonance images. Int Urogynecol J Pelvic Floor Dysfunct. 2007;18(7):773–8.
110. Miller JM, Umek WH, Delancey JO, Ashton-Miller JA. Can women without visible pubococcygeal muscle in MR images still increase urethral closure pressures? Am J Obstet Gynecol. 2004;191(1):171–5.
111. DeLancey JO, Morgan DM, Fenner DE, Kearney R, Guire K, Miller JM, et al. Comparison of levator ani muscle defects and function in women with and without pelvic organ prolapse. Obstet Gynecol. 2007;109(2 Pt 1):295–302.
112. Hanzal E, Berger E, Koelbl H. Levator ani muscle morphology and recurrent genuine stress incontinence. Obstet Gynecol. 1993;81(3):426–9.
113. Vakili B, Zheng YT, Loesch H, Echols KT, Franco N, Chesson RR. Levator contraction strength and genital hiatus as risk factors for recurrent pelvic organ prolapse. Am J Obstet Gynecol. 2005;192(5):1592–8.
114. Cazemier M, Terra MP, Stoker J, de Lange-de Klerk ES, Boeckxstaens GE, Mulder CJ, et al. Atrophy and defects detection of the external anal sphincter: comparison between three-dimensional anal endosonography and endoanal magnetic resonance imaging. Dis Colon Rectum. 2006;49(1):20–7.
115. Dobben AC, Terra MP, Slors JF, Deutekom M, Gerhards MF, Beets-Tan RG, et al. External anal sphincter defects in patients with fecal incontinence: comparison of endoanal MR imaging and endoanal US. Radiology. 2007;242(2):463–71.
116. West RL, Dwarkasing S, Briel JW, Hansen BE, Hussain SM, Schouten WR, et al. Can three-dimensional endoanal ultrasonography detect external anal sphincter atrophy? A comparison with endoanal magnetic resonance imaging. Int J Colorectal Dis. 2005;20(4):328–33.
117. Pinta T, Kylanpaa ML, Luukkonen P, Tapani E, Kivisaari A, Kivisaari L. Anal incontinence: diagnosis by endoanal US or endovaginal MRI. Eur Radiol. 2004;14(8):1472–7.
118. Burhenne HJ. Intestinal evacuation study: a new roentgenologic technique. Radiol Clin. 1964;33:79–84.
119. Hock D, Lombard R, Jehaes C, Markiewicz S, Penders L, Fontaine F, et al. Colpocystodefecography. Dis Colon Rectum. 1993;36(11):1015–21.
120. Kelvin FM, Maglinte DD, Benson JT, Brubaker LP, Smith C. Dynamic cystoproctography: a technique for assessing disorders of the pelvic floor in women. AJR Am J Roentgenol. 1994;163(2):368–70.
121. Halligan S, Spence-Jones C, Kamm MA, Bartram CI. Dynamic cystoproctography and physiological testing in women with urinary stress incontinence and urogenital prolapse. Clin Radiol. 1996;51(11):785–90.
122. Taylor SA, Halligan S. Imaging techniques. In: Stoker J, Taylor SA, Delancey JOL, editors. Imaging pelvic floor disorders. Berlin: Springer; 2008.
123. Halligan S, Bartram CI, Park HJ, Kamm MA. Proctographic features of anismus. Radiology. 1995;197(3):679–82.
124. Rex DK, Lappas JC. Combined anorectal manometry and defecography in 50 consecutive adults with fecal incontinence. Dis Colon Rectum. 1992;35(11):1040–5.
125. Hiltunen KM, Kolehmainen H, Matikainen M. Does defecography help in diagnosis and clinical decision-making in defecation disorders? Abdom Imaging. 1994;19(4):355–8.
126. Hetzer FH, Andreisek G, Tsagari C, Sahrbacher U, Weishaupt D. MR defecography in patients with fecal incontinence: imaging findings and their effect on surgical management. Radiology. 2006;240(2):449–57.
127. Collinson R, Cunningham C, D'Costa H, Lindsey I. Rectal intussusception and unexplained faecal incontinence: findings of a proctographic study. Colorectal Dis. 2009;11(1):77–83.

Surgical Treatments for Incontinence

12

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Introduction

Faecal incontinence results from an impaired ability to control gas or stool to allow evacuation at a socially acceptable time and place. Normal continence depends on the consistency of the stool, capacity of the rectum, anorectal sampling reflex, and normal resting anal tone. It is maintained by the integrated action of the anal sphincters, the pelvic floor muscles, and intact neural pathways. Incontinence may result whenever any of these mechanisms malfunction without adequate compensation. Treatment for faecal incontinence can be either medical or surgical. The aim of medical therapy is to alter stool consistency through dietary changes and antidiarrhoeal medications, with a concurrent or subsequent course of biofeedback. The multiple surgical alternatives range from minimally invasive procedures, such as injection of bulking agents and sacral nerve stimulation, to complete replacement of the sphincter mechanism with an artificial bowel sphincter or stimulated graciloplasty. We group currently available surgical alternatives into five categories: Repair, Augmentation, Replacement, Stimulation and Diversion/Bypass. However, despite the plethora of exciting advances, a stoma may be the most suitable option for certain patients.

Conservative Management

Conservative medical management is the initial therapy for faecal incontinence. Even when a surgical procedure is being contemplated, it is important to begin treatment with conservative management; this approach helps to optimize the outcomes of the impending procedure. Conservative management focuses on stool consistency. Thus, the goal is to deliver a soft, well-formed stool bolus to the rectum. The Bristol Stool Chart is useful for helping patients understand the different degrees of stool consistency. The chart can also be used to evaluate and guide therapies [1]. The validated Wexner Incontinence score is widely used for assessment of continence,

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with a score of 0 for perfect continence and 20 for complete incontinence [2].

Patients with faecal incontinence commonly have diarrhoea or loose bowel movements. In a study of older adults, 50 % of patients with chronic diarrhoea also had faecal incontinence [3]. Patients with diarrhoea should be evaluated for infectious and inflammatory causes, malabsorption, and endocrinopathies such as diabetes and hyperthyroidism. A full discussion of diarrhoea and its causes is beyond the scope of this chapter. If treating the underlying cause of diarrhoea and ceasing all laxative abuse does not improve the patient's continence, treatment can begin with bulking and anti-motility agents. The most commonly used bulking agents are natural and synthetic fibre. Soluble fibres include psyllium and gum arabic. Insoluble fibres include methyl cellulose and the synthetic calcium polycarbophil. An open-label randomized trial conducted by Bliss et al. demonstrated that, compared with a placebo group, patients with loose or liquid stools who increased their fibre intake experienced a 50 % reduction in the number of incontinent episodes [4]. Fibre has also been shown to increase stool frequency in patients with constipation, which contributes to faecal incontinence by creating overflow.

The most commonly used anti-motility agents are loperamide, codeine, tincture of opium, diphenoxylate plus atropine, and amitriptyline. Clinical trials in patients with diarrhoea and faecal incontinence demonstrate that more patients who are treated with these agents achieve full continence than those treated with placebo [5]. Loperamide, codeine, and tincture of opium are opioids that exert their effects via opioid receptors in the bowel. Diphenoxylate plus atropine and amitriptyline exerts their effects via anticholinergic pathways. Elderly patients may not tolerate the anticholinergic side effects of these medications; thus, caution should be taken accordingly. In a small double-blinded crossover trial of loperamide, codeine, and diphenoxylate, the authors concluded that loperamide was at least as effective as, or better than, the other two agents and had fewer side effects [6].

Diet therapy usually consists of increasing fibre and water intake and avoiding caffeine and alcohol. Patients are advised to consume 20–35 g of fibre and 8–10 glasses of water per day. This regimen is difficult to follow for most patients, and many will require a fibre supplement to achieve >20 g of daily fibre. Caffeine, alcohol, and certain foods may cause diarrhoea, leading to worsening of incontinence symptoms.

Repair Sphincteroplasty

Repair of a sphincter injury due to obstetric, iatrogenic, or other traumatic causes is the most traditional and widely available therapy for faecal incontinence. Obstetric injury is the most common indication for sphincteroplasty. Primary repair at the time of vaginal delivery is almost exclusively performed by obstetricians. Midline episiotomy, prolonged second stage of labour, and a forceps delivery increase the risk of sphincter injury. Studies of women evaluated with endoanal ultrasound after vaginal delivery have revealed sphincter defects in 26.9 % of primiparous women and new sphincter defects in 8.5 % of multiparous women [7]. Approximately 30 % of women with a sphincter defect will have faecal incontinence.

Endoanal ultrasound remains the primary tool for identification of sphincter injuries. The role of preoperative physiologic testing remains controversial. Gilliland et al. reviewed the outcomes of 77 patients who underwent preoperative physiologic testing followed by anterior overlapping sphincteroplasty [8]. Age, parity, prior sphincteroplasty, duration of incontinence, size of defect on endoanal ultrasound, and manometric parameters did not correlate with outcomes. Seventy-one patients underwent preoperative pudendal nerve terminal motor latency (PNTML) testing. Among patients with either a unilateral or bilateral pudendal neuropathy, only 16.7 % had a successful outcome compared with 62 % of patients who had a normal study. The authors concluded that all patients with faecal incontinence and an external sphincter defect should be offered anterior overlapping sphincteroplasty. A further recommendation was that patients should be informed that the integrity of the pudendal nerves is the most important predictor of success.

Goetz and Lowry identified 16 papers in which the influence of pudendal neuropathy was assessed [9]. Five papers concluded that neuropathy predicted outcomes after sphincteroplasty. In contrast, 11 studies that included more than 700 patients reported no such relationship. The authors

concluded that PNTML does not predict postoperative function and should not be used to exclude patients from surgery; clearly no consensus exists.

It is generally agreed that unless repair is attempted immediately after injury, it should be delayed until the wound has healed and inflammation has subsided. Studies on primary overlapping sphincter repair from the gynaecology literature have had mixed results [10].

Preoperatively, all patients who undergo sphincteroplasty are given a bowel prep of polyethylene glycol. Patients receive preoperative antibiotics in accordance with guidelines of the National Surgical Quality Improvement Program (NSQIP) of the American College of Surgeons. After general anaesthesia, the patient is placed in the prone jackknife position. The buttocks are separated with tape, and the anus and perineum are prepped with a povidone iodine solution. A transverse curvilinear incision is made overlying the external anal sphincter, approximately 0.5 cm caudal from the anal verge. The external sphincter is then dissected both from the internal anal sphincter and the posterior wall of the vagina. The anterior scar is sharply divided, but not excised. Care is taken to preserve the scar, which helps to hold the sutures. Interrupted long-term absorbable sutures are placed along the internal anal sphincter to plicate the sphincter. The desired effect is to place an index finger through a snug, but not tight, repair. An anterior levatorplasty can also be undertaken at this point. The ends of the external sphincter are overlapped and sutured to each other with a series of interrupted absorbable sutures in a mattress fashion. The wound is then partially closed in a Y-fashion to further separate the anus from the vagina. The central portion of the wound should be left open. It is not necessary to routinely use faecal diversion or bowel confinement [11, 12].

Studies with less than 3 years of follow-up have demonstrated promising results for anterior overlapping sphincteroplasty. In 55 patients treated with overlapping sphincteroplasty, Fleshman et al. reported that 28 (51 %) had complete continence and 12 (22 %) were only incontinent to gas at 1 year of follow-up [12]. Similarly, Wexner et al. found that in 16 patients with a mean follow-up of 10 months, 72 % reported excellent or good functional results postoperatively [13]. Engel et al. performed ultrasound and physiology testing pre- and postoperatively [15]. At a median of 15 months of follow-up, they found that 76 % of patients had improvement after the repair. These researchers also observed that a larger increase in squeeze pressure (20 vs 5 cm H_2O, $p=0.05$) and an intact external anal sphincter after repair (32 of 35 vs 5 of 11, $p=0.003$) correlated with success.

Results of overlapping sphincteroplasty in the long term are far less encouraging. Halverson and Hull reviewed the experience from the Cleveland Clinic [16]. With a median follow-up of 62.5 months, 49 patients were contacted and Fecal Incontinence Severity Index (FISI) and Fecal Incontinence Quality of Life Scale (FIQL) scores were calculated. Thirty-one patients had an injury due to obstetric causes, 7 were iatrogenic, 3 were from trauma, and 3 were not documented. Four patients had subsequent permanent faecal diversion after repair. More than 50 % of the patients were incontinent to liquid and solid stool, and only 14 % reported perfect continence. The median patient-rated and surgeon-rated FISI score was 20 [16]. This group of researchers published a subsequent study with a median follow-up of nearly 11 years [17]. At the study end point, none of the patients reported perfect continence, and the patient-rated and surgeon-rated FISI score increased to 39.4 and 39.9, respectively. These results indicate a significant decline in function as compared with the prior study.

Postanal Repair

Sir Alan Parks developed the posterior anal repair, which now bears his name, in the late 1960s and early 1970s. The repair was designed for patients with idiopathic faecal incontinence with the intention to restore the acute angulation of the anorectal junction. The repair initially showed promising results through the 1980s with reported success rates up to 86 % [18]. The results of these early studies may have been limited by a lack of standardization in grading of faecal incontinence. With the advent of anorectal ultrasound, the identification of anterior sphincter injuries became more common, and it is postulated that some of the patients initially treated with a posterior repair for "idiopathic" faecal incontinence indeed likely

had an anterior sphincter defect. Matsuoka et al. reported the results of 21 patients who underwent posterior repair after thorough preoperative evaluation [19]. None of the patients had a sphincter defect and 13 had prolonged pudendal nerve terminal latencies. Thirty-five percent of patients reported improvement in their symptoms. In this group, the Wexner faecal incontinence score improved from a mean of 16.5 preoperatively to 2.6 postoperatively, which was statistically significant. The remaining patients who reported no improvement had a minimal change in their incontinence score, from 16.5 preoperatively to 13.3 postoperatively (not statistically significant). The most recent series, reported by Mackey et al., evaluated 57 patients [20]. Postoperative incontinence was rated by patients as *none* to *minimal* (26 %), moderate (26 %), and severe (48 %).

Technique

The Parks' postanal repair is performed with patients under general anaesthesia in the prone jackknife position. A posterior angulated or curvilinear incision is made 1–2 cm distal to the anal verge. An intersphincteric dissection is then undertaken to above the level of the levators and into the presacral space. The ischiococcygeus and puborectalis are then approximated in layers with interrupted absorbable suture. The external sphincter is similarly plicated, and a finger is introduced into the anal canal to assess the repair. The wound is closed in layers, often over a closed suction drain.

Augmentation Radiofrequency Therapy

The Secca® procedure, which was approved by the US Food and Drug Administration (FDA) for treating faecal incontinence, involves the delivery of radiofrequency energy to the anal canal. The basis for such therapy comes from experience in applying radiofrequency energy to the lower oesophageal sphincter in treating gastroesophageal reflux disease. The Mederi RF generator (Mederi Therapeutics, Inc., Greenwich, CT) delivers radiofrequency energy at 465 kHz to the muscle below the mucosa through a set of four needles (Fig. 12.1). The power output is varied to achieve a target temperature of 85 °C. The mucosa is protected with water irrigation. The lesions are delivered from 1.5 cm above the dentate line to 0.5 cm below the dentate line. 16 to 32 applications are delivered, with each set comprising 4 individual lesions.

The results of various series of the Secca® procedure reported in the literature are summarized in Table 12.1. The first reported experience with the Secca® procedure was by Takahashi et al. in 2002 [21]. In a pilot study of 10 patients with 12 months of follow-up, these researchers demonstrated a significant reduction in Wexner faecal incontinence scores, from 13.5 to 5.0. At 24 months of follow-up, the average Wexner score in this same cohort was 7.8 [23]. This was still a significant decrease from baseline but not as large as that seen at 1-year of follow-up. A multicenter, prospective, manufacturer-sponsored study in the United States conducted by Efron and colleagues included 50 patients who were followed for 6 months. The results of this study demonstrated a more modest decrease in Wexner scores from 14.5 to 11.1 [22]. However, the authors noted an improvement in all four components of the FIQL score as compared with baseline. Ruiz et al. noted a similar modest decrease in Wexner scores from 15.6 to 12.9 at 12 months in patients treated with the Secca® procedure [26]. Lefebure et al. reported a minimal change in Wexner scores in a series of patients followed for 12 months [25].

Because the device was unavailable for several years, recent data are limited. However, the Secca® procedure remains a viable option. It is a minimally invasive procedure that can be performed under local anaesthesia, and it has few associated complications. In patients with incontinence who do not have a known sphincter defect, the number of surgical options is limited. Further studies may help elucidate which of these patients may benefit most from the Secca® procedure.

Injectables

The symmetry and anatomy of the anal canal have been recognized as important components in the maintenance of continence. The use of injectable

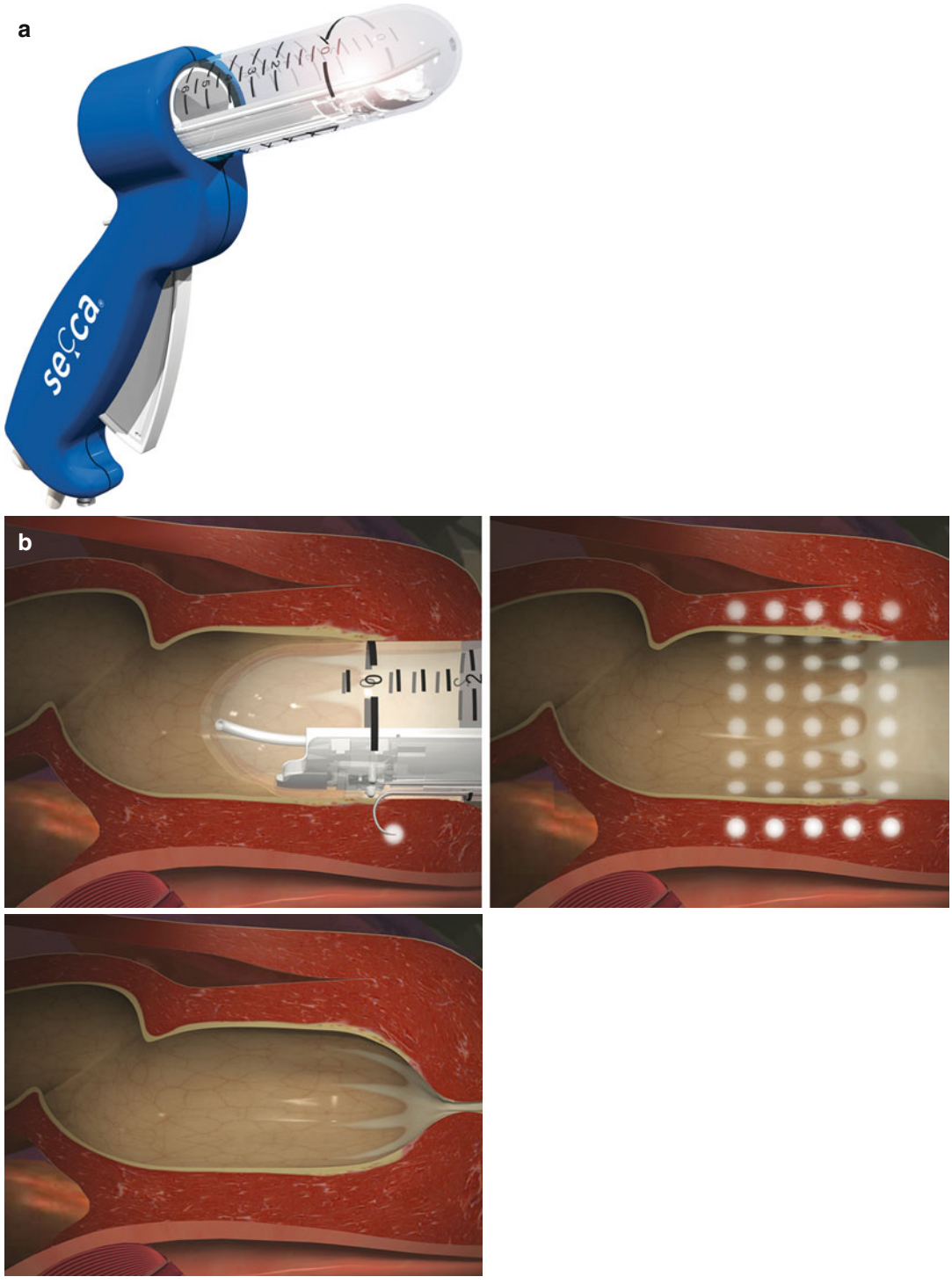

Fig. 12.1 Schematic illustration of (**a**) the applicator, (**b**) four quadrant electrode deployment sites (© 2012 Mederi Therapeutics Inc.)

Table 12.1 Summary of trials on radiofrequency energy therapy (Secca® procedure)

Author (year)	*n*	Follow-up (months)	Wexner score		Comments
			Before	After	
Takahashi (2002) [20]	10	12	13.5	5	Four bleeding complications, 3 resolved, 1 required suture control
Efron (2003) [21]	50	6	14.5	11.1	18 % had prior overlapping sphincteroplasty; 4 % had prior artificial bowel sphincter
Takahashi (2003) [22]	10	24	13.7	7.8	No significant difference between 1- and 2-year follow-up
Takahashi (2008) [23]	19	60	14.4	8	Sustained benefit
Lefebure (2008) [24]	15	12	14.1	12.3	No long-term complications. No change in FIQL scores except in the depression subscale
Ruiz (2010) [25]	16	12	15.6	12	No long-term complications

Table 12.2 Summary of trials on injectable materials

Author (year)	*n*	Material used	Follow-up (months)	Wexner score		*P* value
				Before	After	
Shafik (1993) [26]	11	PTFE	24	63 % improved		–
Shafik (1995) [27]	14	Autologous fat	24	85 % improved		–
Malouf (2001) [28]	10	Bioplastique®	6	30 % improved		_
Davis (2003) [29]	18	Durasphere®	28.5	11.8	8	0.002
Tjandra (2004) [30]	82	Silicone (US guided)	12	14.5	3	<0.001
Chan (2006) [31]	7	PTQ®	14	9–14	1–5	0.016
Stojkovic (2006) [32]	73	Contigen®	12	10	6	<0.001
de la Portilla (2008) [33]	20	PTQ®	24	13.5	9.4	0.127
Maeda (2008) [34]	10	Bulkamid®	19	15	12	<0.05
Maeda (2008) [34]	10	Permacol®	19	16	15	<0.05
Soerensen (2009) [35]	33	Silicone	12	13	11	–
Tjandra (2009) [36]	20	PTQ®	12	12	4	<0.001
Graf (2011) [37]	206	NASHA/Dx	12	14.3	10.9	<0.001

bulking agents for the treatment of faecal incontinence stems from similar technology in the field of urinary incontinence. Table 12.2 lists several of the early and more recent trials on injectable agents. Use of these agents is attractive due to their minimally invasive nature. First described for faecal incontinence by Shafik and colleagues in 1993, submucosal injection of polytetrafluoroethylene (Teflon or Polytef™; DuPont, Wilmington, NE) and autologous fat yielded successful short-term outcomes. Other injected agents include carbon-coated beads, silicone, collagen, nonanimal stabilized hyaluronic acid stabilized in dextranomer microspheres, polyacrylamide, and porcine dermal collagen.

The Cochrane Collaboration reviewed the available literature on injectable agents in 2010. Based on four randomized trials involving 176 patients, the authors observed a high risk for bias, which precluded definitive conclusions [39]. They did note, however, that most trials showed an improvement in patients' symptoms in short-term follow-up. A small randomized trial comparing silicone biomaterial and carbon-coated beads showed a greater proportion of patients in the silicone group having a 50 % reduction in incontinent episodes at 12 months of follow-up [37]. The injection method employed in each of the reported studies is highly variable. In a study including 82 patients who underwent injection

of silicone, Tjandra et al. reported significantly improved functional and quality of life outcomes with ultrasound-guided injections compared with non-guided injections [31].

Noting the relative lack of high-quality, randomized data, the NASHA Dx Study Group conducted a manufacturer-sponsored, international, multicenter, randomized, double-blinded controlled trial at 8 US centres and 5 European centres [38]. Patients with a Wexner score of greater than 10 and who had failed conservative management were randomized in a 2:1 fashion to transanal submucosal injection with nonanimal stabilized hyaluronic acid stabilized in dextranomer microspheres (Solesta®, Salix Pharmaceuticals, Raleigh, NC) or sham therapy, which consisted of mimicking the procedure without injection of any substance. The primary end point was a response to treatment as defined by a reduced number of incontinent episodes by 50 % or more compared with baseline. Fifty-two percent of patients in the treatment group achieved this end point at 6 months compared with 31 % of patients in the sham arm (odds ratio 2.36, 95 % CI 1.24–4.47, $p=0.009$). There was no difference in the median decrease in number of incontinent episodes or change in Wexner scores from baseline between the treatment and sham groups. There was a significant difference between the treatment and sham groups in the mean increase in number of incontinence-free days at 6 months (3.1 vs 1.7, $p=0.016$) [38].

The role of injectable materials for faecal incontinence has not yet been fully defined. However, their demonstrated efficacy, minimally invasive nature, and low complication rates are certainly advantageous attributes that may support the use of this therapy, either as a primary treatment of faecal incontinence or as an adjunct. The long-term results, optimal dose, and site of injection are all issues that remain to be clarified.

Replacement Muscle Transposition: Non-stimulated Gluteoplasty

Gluteus maximus muscle transposition was described in the early twentieth century as a treatment for faecal incontinence [40]. The gluteus maximus muscle is suitable for transposition due to its large bulk, proximity to the anal canal, and single proximal neurovascular pedicle. In addition, buttock contraction is a standard response to impending incontinence [41].

Technique

In the prone jackknife position, the lower portion of the gluteus maximus muscles and the fascia are individually mobilized from their origins on the ileum and sacrum. The neurovascular bundle is identified near the ischial tuberosity and preserved. The muscle strips are tunnelled underneath the skin and anchored to the contralateral gluteus maximus muscle on each side so that the anus is fully encircled [41].

Outcomes

Devesa et al. reported the largest series of bilateral gluteoplasties for faecal incontinence, in which 9 of 17 patients achieved normal control and the most common reported morbidity was infection of the perianal wound [42]. However, with the introduction of gracilis transpositions, enthusiasm for the gluteoplasty diminished [41].

Muscle Transposition: Non-stimulated Graciloplasty

In 1952, Pickrell described a novel surgical approach to treating children with faecal incontinence caused by neurologic and congenital disorders [43]. The advantages of transposition of the gracilis muscle include its superficial location, ease of mobilization, and lack of requirement for strength or range of motion [41].

Technique

The technique entailed removing the gracilis muscle from the thigh, wrapping it around the anus, and attaching the free end to the contralateral ischial

tuberosity. Patients underwent training to gain voluntary control of muscle contraction and relaxation.

Outcomes

Corman reported long-term outcomes of non-stimulated graciloplasty; 11 of 14 patients in this study had fair to excellent results [44]. Christiansen et al. reported a series of 16 patients who underwent unstimulated gracilis transpositions, with over 80 % improvement [45]. In an attempt to improve outcomes, Kumar et al. performed bilateral gracilis transpositions in 10 patients. They observed a 90 % improvement in continence maintained for 2 years [46].

Muscle Transposition: Stimulated Graciloplasty

The non-stimulated graciloplasty technique suffered from faced with certain long-term limitations. Namely, the chronic contraction of the transplanted gracilis muscle caused fatigue and thereby compromised the patient's sustained control of continence. The gracilis muscle is naturally composed of fast-twitch type II fibres that are easily fatigable. An approach to resolving this limitation became available in the early 1980s, when researchers demonstrated that the application of low-frequency electrical stimulation could, over time, transform type II fibres into slow-twitch, fatigue-resistant type I fibres [47]. In 1988, Baeten and coworkers used this technology to advance the procedure of stimulated graciloplasty. Their approach, which is also called dynamic graciloplasty, involved stimulating the gracilis muscle with a pulse generator. Tested on patients with fatigue-related suboptimal control following transposition, this stimulation was demonstrated to engender a neosphincter characterized by involuntary resting tone [41, 48].

Technique

Depending on whether a diverting stoma is created, the procedure of stimulated graciloplasty is carried out in two or three phases. In the first phase, the gracilis is removed from the thigh and wrapped around the anus to form a skeletal muscle ring; the severed distal portion is anchored to the contralateral ischial tuberosity (Fig. 12.2). The stimulator (Medtronic Inc, Minneapolis, MN) is implanted in the abdominal wall (Fig. 12.2c), with leads placed on the main trunk or in the intramuscular portion of the gracilis muscle, close to the nerve. The second phase of the procedure involves conditioning the muscle through the application of low-frequency neuromuscular stimulation, delivered at increasing levels over a period of 8 weeks. This process gradually changes the contractile properties of the gracilis muscle, transforming its easily fatigable fast-twitch fibres into fatigue-resistant slow-twitch fibres. In this phase, the patient also learns to use an external magnet in order to turn the stimulator on and off, thereby causing muscle contraction and relaxation, which facilitates the control of continence [41, 49]. If a diverting stoma must be created, additional operative interventions are required for its creation and subsequent closure.

Outcomes

Stimulated graciloplasty was a widely applied transposition procedure for treating faecal incontinence. Success rates generally range between 57 and 93 % [41, 49]. As reported in a prospective series including 17 patients, our initial experience with the procedure at Cleveland Clinic Florida indicated its feasibility despite a steep learning curve [50]. In an initial report of a multicenter trial, the Dynamic Graciloplasty Therapy Study Group (DGTSG) found that 60 % of patients who underwent the procedure had significant improvements in continence and quality of life [51]. Wexner et al. reported the long-term results of this multicenter trial, including outcomes of 129 patients who underwent stimulated graciloplasty from 1993 to 1999 [52]. Overall success, defined as a 50 % or greater reduction in the number of faecal incontinence episodes, was achieved in 62 and 56 % of patients at 1 and 2 years, respectively. These rates demonstrate the durability of the earlier, short-term DGTSG results. The authors of a systematic review

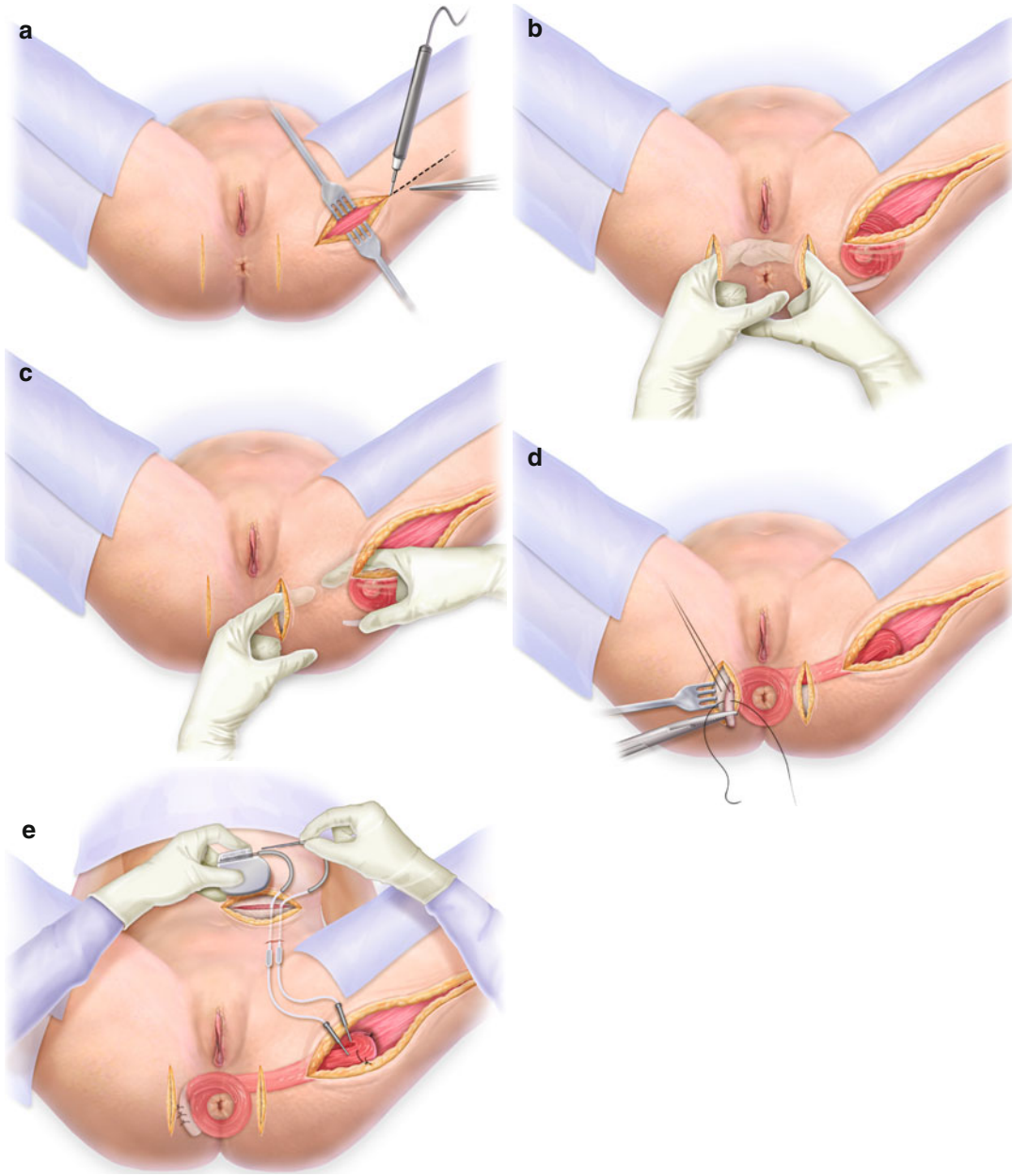

Fig. 12.2 (**a**) Incisions for harvesting the gracilis muscle. (**b**) Incision at the anus. (**c**) Incisions for tunneling the gracilis muscle from the leg to the anus. (**d**) Wrapping the gracilis muscle around the anus. (**e**) The stimulator that is implanted in the abdominal wall. With permission from Wolters Kluwer copyright 2012

reported that the efficacy of stimulated graciloplasty, as measured by patient reports of satisfactory continence, ranged from 42 to 85 % [53].

Many series have demonstrated that stimulated graciloplasty is effective in treating faecal incontinence; however, relatively high rates of complications and surgical revision have also been observed (Table 12.3). As reported by the DGTSG in the original trial, rates of complications and reoperations were 74 and 40 %, respectively [41, 57]. We observed a variety of complications associated with this procedure and its associated technology, including lead fibrosis, seroma of the thigh incision, excoriation of the

Table 12.3 Outcomes of stimulated graciloplasty

Author (year)	*n*	Follow-up (months)	Morbidity (%)	Revision surgery (%)	Success (%)
Christiansen (1998) [53]	13	17	–	–	84
Sielezneff (1999) [54]	16	20	50	43.7	81
Mavrantonis (1999) [55]	21 IM	21	–	–	93
	6 DS	12.5			10
Mander (1999) [56]	64	10	–	–	56
Madoff (1999) [57]	128	26	41	–	66
Konsten (2001) [58]	200 IM	–	–	2.7	74
	81 DS			26	57
Bresler (2002) [59]	24	–	42	46	79
Wexner (2002) [51]	129	24	–	–	62
Rongen (2003) [60]	200	72	–	69	72
Penninckx (2004) [61]	60	53	77	77	61

IM intramuscular, *DS* direct stimulation

skin above the stimulator, rotation of the stimulator, premature battery discharge, fracture of the lead, perineal skin irritation, perineal sepsis, rupture of the tendon, tendon erosion, muscle fatigue during programming sessions, electrode displacement from the nerve, and fibrosis around the nerve [50]. In addition, some patients who underwent the procedure had faecal impaction, anal fissure, and parastomal hernia. Other studies have reported instances of hardware failures, infections, muscle detachment, device malfunction and migration, postoperative evacuatory dysfunction, and severe unresolved pain that resulted in hospitalization and reoperation in numerous cases [53]. Some of these complications led to stoma creation or death.

In 93 cases of dynamic graciloplasty, Matzel et al. reported 211 complications [63]. Although 42 % of patients in this study had severe complications, 92 % achieved recovery following treatment. With the exception of major infections, most of the complications did not adversely affect outcomes. In a systematic review of adverse events associated with the procedure, the most common complications observed were infection (28 %), device malfunction (15 %), and leg pain (13 %) [49, 53]. The mean morbidity rate was 1.12 per patient (range 0.14–2.08).

Risks of morbidity associated with stimulated graciloplasty have been reduced through selected modifications of the procedure. For example, rates of infectious complications have been lowered as a result of improved infection control measures [41, 61]. Complications caused by nerve fibrosis, lead displacement, and high impedance have been virtually eliminated by placing the leads adjacent to the intramuscular portion of the nerve rather than directly on the exposed portion of the nerve trunk [59, 61]. This method of stimulation is the only factor that has been identified as a significant predictor of successful outcomes of stimulated graciloplasty [59]. However, it is evident that surgical experience strongly impacts outcome [41, 58].

Despite evidence for the positive influence of stimulated graciloplasty on patient function and quality of life, Medtronic Inc. abandoned pursuit of FDA approval for the neurostimulator in the United States in 1999. This unfortunate decision was attributed partly to the relatively high rates of associated morbidity. Currently, surgeons in many other countries perform the operation to treat faecal incontinence. In addition, the technique of stimulated graciloplasty has been adapted for performing total anorectal reconstruction for anal atresia and following abdominoperineal resection. In the United States, sphincter muscle loss is currently treated with unstimulated graciloplasty. In the era of FDA approval for sacral nerve stimulation (SNS), graciloplasty still has a role in the treatment of

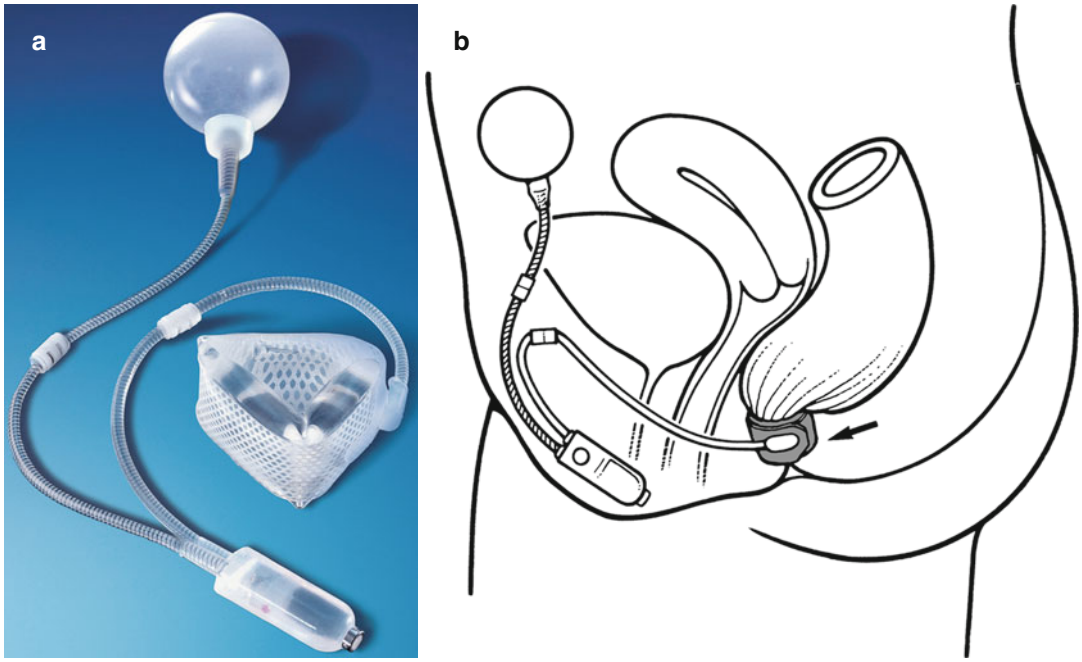

Fig. 12.3 (**a**) Artificial bowel sphincter system, (**b**) schematic of ABS placement for faecal incontinence in a female patient (Courtesy of American Medical Systems, Inc.)

faecal incontinence secondary to large sphincter defects or in patients with rectoanal atresia who are not likely to benefit from SNS and will continue to depend on sphincter replacement. Perhaps successful outcomes for this subgroup of patients might be best achieved with an artificial bowel sphincter preceded by a nonstimulated graciloplasty.

Artificial Bowel Sphincter

This approach to treating faecal incontinence involves implanting a prosthesis that simulates normal anal sphincter function. The Acticon® Neosphincter (American Medical Systems, Minnetonka, MN), which was approved by the FDA in 2001, is an implantable, fluid-filled artificial bowel sphincter (ABS) made of solid silicone rubber. The prosthesis comprises three parts that are connected with kink-resistant tubing: a perianal occlusive cuff, implanted around the anal canal; a pressure-regulating balloon, implanted in the abdomen; and a control pump with a septum, implanted in the labium majus or scrotum (Fig. 12.3) [31, 64, 65]. In response to the patient's control of the pump mechanism, the Acticon® Neosphincter facilitates the opening and closing of the anal canal, simulating normal anal sphincter function. Continence is maintained as adjustments of the fluid-filled cuff compress the anal canal to a pressure approximating physiological resting values.

In order to evacuate, the patient squeezes and releases the pump mechanism between 5 and 15 times. Each pump moves fluid from the cuff to the pressure-regulating balloon, thereby emptying and collapsing the cuff, which releases the compressive force around the anal canal. Due to residual pressure within the balloon, fluid passively flows back into the cuff, usually refilling it within 2–3 min. Continence is re-established as the refilled cuff compresses the anal canal again. Pressure in the occlusive cuff is maintained by the pressure-regulating balloon. The ABS device is available in different cuff lengths (8–14 cm), widths (2.0–2.9 cm), and balloon pressure ranges (80–120 cm H_2O).

Table 12.4 Outcomes of artificial bowel sphincters

Author (year)	*n*	Follow-up (months)	Infection (%)	Device explant/reimplant	Functional (%)
Wong (1996) [65]	12	58	25	7/4	75
Lehur (1998) [66]	13	30	8	4/2	85
Vaizey (1998) [67]	6	10	33	1/0	83
Christiansen (1999) [68]	17	60	18	7/0	53
Lehur (2000) [69]	24	20	4	8/4	83
O'Brien (2000) [70]	13	–	23	3/0	77
Altomare (2001) [71]	28	19	18	5/0	75
Lehur (2002) [72]	16	25	0	6/1	75
Devesa (2002) [73]	53	26.5	21	12/2	49
Ortiz (2002) [74]	22	28	9	9/2	68
Wong (2002) [75]	112	12	38	41/7	67
Michot (2003) [76]	25	34.1	12	5/0	76
Parker (2003) [77]	37	12	19	27/7	49
Casal (2004) [78]	10	29	10	3/2	90
Ruiz-Carmona (2008) [79]	17	68	29	11/3	53
Wexner (2009) [80]	47	39	41	18/4	65

Technique

Because infection is a major complication of the ABS procedure, meticulous aseptic technique is imperative. Preoperatively, patients undergo a full bowel preparation and receive appropriate intravenous antibiotic prophylaxis. A modified lithotomy position is preferred in order to allow a combined perineal and abdominal approach. An anterolateral circumanal incision in the rectovaginal or rectourethral septum is created, and dissection proceeds in a cephalad direction. The ischiorectal fossae are entered on both sides, and a circumferential tunnel around the rectum is created, ideally proximal to the anococcygeal ligament, to minimize the chance of erosion through the perianal skin. This incision must be made very carefully to avoid injuring either the rectum or the vagina, as injury would preclude implantation of the artificial sphincter. A cuff sizer is passed around the anal canal to allow correct selection of the cuff width and length. The cuff itself is passed around the anus and fastened. A low transverse abdominal incision is made and a pocket in the space of Retzius is created. The cuff tubing is then tunneled up from the perineal incision towards this pocket. The balloon is inserted in the pocket. The control pump is implanted in the subcutaneous tissues of the scrotum in men and the labium majus in women on the ipsilateral side of the patient's dominant hand. The balloon is filled with fluid and connected to the cuff tubing for 30 seconds to allow for pressurization of the cuff. The balloon is then drained and refilled with the appropriate amount of fluid. The final step is implantation and pressurization of the pump, tubing, and cuff via a colour-coded tubing system. The device is kept in the deactivated state until the surgical wounds have healed, generally over a period of 6 weeks after which it is activated in the outpatient setting.

Outcomes

Since Christiansen et al. first reported on ABS as a treatment for faecal incontinence in 1987, numerous case series have been published on the efficacy and safety of the procedure (Table 12.4) [31, 64, 82]. Wong et al. reported a large multicenter, prospective trial including 112 patients [76]. The findings demonstrated that the ABS is effective, achieving a continence rate of 85 % and a significant improvement in quality of life in patients with functioning devices. However, on an intention-to-treat basis, success was achieved in only 53 % of patients.

Similar to stimulated graciloplasty, limitations to the ABS are attributable to its high rate of complications; most of these are related to infections of the foreign material and subsequent need for surgical revision and explantation. A systematic review of ABS case series reported a statistically and clinically significant improvement in Wexner and AMS scores after ABS implantation [83]. Six studies reported quality of life outcomes, which were also significantly improved compared with preoperative assessment. However, preoperative and postoperative functional and quality of life outcomes were not assessed by an intention-to-treat analysis in any of these studies; studies only compared outcomes in patients with a functional device. Thus, any negative impact of implantation followed by explantation of a failed ABS device was not assessed, possibly biasing the results. Ruiz-Carmona et al. reported long-term outcomes of ABS in 17 patients with a median follow-up of 5 years [80]. Only 9 of 17 (53 %) cases had an activated functional ABS by the end of the study period. However, those nine patients had significantly improved continence, with significant improvement in Wexner scores from 17.5 preoperatively to 9, 5.5 and 10 at 6, 12 months and at the end of follow-up, respectively. In addition, there was a significant improvement in quality of life.

Unfortunately, rates of postoperative complications of ABS have remained very high, ranging from 19 to 100 % [31, 64]. A systematic review including 14 studies reported explantation rates between 17 and 41 % and surgical revision rates between 13 and 50 % [83]. Wong et al. reported 117 postoperative complications in 114 cases [76]. The most common complications include infections, erosions or ulcerations of the rectum or surrounding skin, device malfunction such as cuff rupture, balloon and pump leaks, and device migration [76, 80, 81, 83, 84]. Fecal impaction, dehiscence of the perineal wound, pain, discomfort, and patient dissatisfaction are less common but also significant problems [81, 83, 84]. Among these complications, infection is the most common, resulting in explantation in 4–38 % of cases [76, 80, 81, 83, 84]. Ruiz-Carmona reported 5-year follow-up data in 17 patients with an ABS [80]. All patients suffered from at least one complication, and 65 % required at least one reoperation. After the first implant, 11 devices had to be removed (65 %) and 7 patients eventually underwent a second implantation.

ABS infection can be divided into two groups: early-stage infection, defined as infection prior to ABS activation, and late-stage infection, defined as infection after ABS activation [81, 83]. In a study from our institution, 21 of 51 patients (41.2 %) developed infection at a mean follow-up period of 39 months [81]. Eighteen (35.3 %) cases developed infection before ABS activation (early-stage infection), a result similar to other reports; all 18 cases required ABS explantation. A bowel movement prior to the third postoperative day and a history of perianal infection prior to ABS implantation were risk factors for early-stage ABS infection. In a study by Winslett et al., secondary procedures were necessary in up to 32 % of the patients treated for perianal abscess, either because of inadequate examination under anaesthesia for drainage or undefined occult fistula-in-ano [85].

Late-stage complications after ABS device activation can also result in ABS explantation. The incidence of these complications may increase with device use over time [78, 86]. In a series from our institution, the most common late-stage complication was device malfunction, followed by erosion of device, persistent perianal pain, migration of device, constipation, and hematoma over the labium majus [81]. This was similar to the other reports [74, 76, 78, 83, 84, 86]. Thirteen of 33 patients (32.0 %) required ABS explantation. In the late stage, device malfunction was the most common reason for explantation (46.1 %). All of the ABS devices that required explantation functioned satisfactorily after activation, but the function deteriorated with time. Four of 9 malfunctioned ABS devices were due to leakage of the system, which implied that the present design may be inadequate for longer-term function. Erosion of the rectum or skin was the second most common reason for explantation (38.5 %). Five patients had erosions through the skin and one had erosion through the rectum with associated rectovaginal fistula. Five patients had associated infection that resulted in explantation [81]. Tejirian et al. reported intra-abdominal erosion of artificial bowel sphincter reservoir [87].

Devesa et al. attempted to identify risk factors of erosion, but they found no association with pre-existing fibrosis, perianal wound closure method, tension of the wound, and soiling or straining during evacuation [74]. Similarly, we did not find erosion to be associated with any important patient-related factors [81]. Erosions were found only after ABS activation. The rate of explantation increased with the time after ABS implantation; the longer the ABS was in use, more complications occurred and the risk of ABS explantation increased. The 1-year and 2-year cumulative risks of ABS explantation were 9.7 and 13 %, respectively. After 2 years, the risk of ABS explantation sharply increased, and in the third and fourth year, risk increased to 43 and 48 %, respectively [81]. A similar explantation rate of 44 % at 48 months was also reported by Ortiz et al. [75].

Despite these shortcomings, ABS is an effective method of achieving continence in patients with debilitating faecal incontinence who might otherwise require a permanent colostomy. It certainly requires a motivated, healthy patient who is technically able to operate the device and is willing to potentially undergo multiple operations in order to achieve continence.

Stimulation: Sacral Nerve Stimulation

Initially developed in 1989 to treat urinary incontinence, SNS is a promising approach for patients with faecal incontinence. In 1995, Matzel et al. reported initial success with this treatment for faecal incontinence in three patients [88]. Since that time, extensive experience with SNS has been gained in Europe and Australia, and the technique has been refined [89, 90]. Most recently, after a large multicenter trial [91], this procedure was FDA approved for faecal incontinence in the United States.

Technique

We evaluate all SNS candidates by a physiological assessment and by obtaining a Wexner faecal incontinence score, in addition to completing a 2-week bowel diary. SNS is performed in two stages. The first stage involves the placement of a tined lead in the third sacral foramen under fluoroscopic guidance. The patient is awake for the beginning of this procedure but is given intravenous sedation and local anaesthesia as needed. Once the needle is placed in the third sacral foramen, the patient's motor and sensory response is tested by connecting the needle to a temporary pulse generator. A successful sensory response includes any sensation (such as tapping or tightening) in the perineum, vagina, scrotum, rectum or pelvis, and the desired motor response is plantar flexion of the great toe and a bellows response in the perineum. Once both responses are elicited, the tined lead is placed using a Seldinger technique with fluoroscopic guidance (Fig. 12.4). Once again, fluoroscopic images are obtained, and sensory and motor testing are performed to ensure proper positioning. Low-voltage stimulation is desired in order to achieve the longest possible battery life. A subcutaneous pocket is then created in a preoperatively marked location in the upper buttock, and the quadripolar lead is attached to a temporary extension that is tunneled into this pocket. The temporary extension is attached to an external pulse generator for the duration of the 2-week test period. The patient is monitored for 2 weeks with a re-evaluation of the Wexner score and maintenance of a bowel diary. Assuming a 50 % or greater improvement in symptoms, the patient returns after 2 weeks for implantation of the permanent stimulator. Because a sensory response is not elicited during this second stage operation, a deep intravenous sedation may be employed. The incision is reopened, the temporary connection is removed, and the lead is placed into the stimulator.

Mechanism of Action

Despite the wide acceptance and application of SNS, the exact mechanisms of action are poorly understood. Fecal continence requires a complex interaction of the puborectalis muscle, internal and external anal sphincter muscles, colonic motility, and anorectal sensory function. During SNS, the

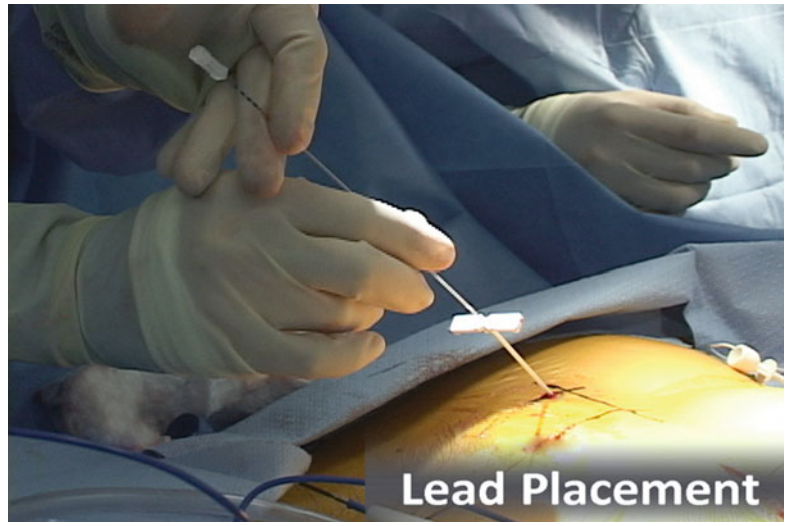

Fig. 12.4 Tined lead placement under fluoroscopic guidance in sacral nerve stimulation stage I. Courtesy of Medtronic

third sacral nerve, a mixed somatic motor and sensory nerve, is stimulated. The stimulation current is rarely set at a level that would induce external anal sphincter contraction, as this would result in continual discomfort; but rather the degree of stimulation is determined by patient sensation of the stimulation in the perineum [92]. It is therefore likely that stimulation of somatic sensory efferent nerves is an important component of the physiological effects. [92] In an animal model, Griffin et al. observed augmented sensory cortical evoked potentials from the anal canal, in addition to upregulation of cortical neural cell adhesion molecule (NCAM) expression with SNS [92, 93]. Sheldon et al. demonstrated cerebral cortical changes with temporary SNS [94]. Goonertane et al. showed normalization of mucosal substance P levels in patients who responded to stimulation [95]. Interestingly, no specific changes in anorectal physiology testing parameters have been consistently proven to be altered by SNS.

A recent prospective study from the United Kingdom included 23 patients who underwent temporary SNS, including 16 patients (70 %) who had a good clinical response to test stimulation [96]. Maximal squeeze pressure increased in all patients; however, resting pressures significantly improved only in responders. SNS did not influence rectal compliance in both responders and nonresponders. Maximal tolerated volumes were significantly increased in all patients after test stimulation; however, maximal tolerated distension significantly increased in responders only.

Otto et al. hypothesized that SNS leads to pelvic floor contraction and increased rectal perception [97]. In a prospective series of 14 patients with consecutively implanted permanent stimulators, these researchers performed endoanal ultrasound, manometry, and volumetry examinations at 6 months with the device on or off. The stimulator settings were placed at subsensory levels 1 month prior to examination so that patients were blinded to the stimulator settings (on/off) at the time of examination. Stimulator activation was associated with decreases in the diameter of the external anal sphincter (8.7–7.0 mm), the diameter of the internal sphincter (3.3–2.7 mm), and the distance between the pubic symphysis and anal mucosa (46.5–43.4 mm). Perception threshold increased from 62.1 to 86.4 mL, and the volume evoking defecation increased from 148.9 to 188.2 mL with the stimulator turned on. However, the intrarectal pressures and the corresponding volumes did not differ with the stimulator on or off [97].

Outcomes

High success rates for test stimulation and permanent SNS implantation have been reported in multiple series (Table 12.5). In a large multicenter,

Table 12.5 Outcomes of sacral nerve stimulation

Author (year)	*n*	Follow-up (months)	Scoring method	Outcome		*P* value
				Before	After	
Malouf (2000) [97]	5	16	Wexner	16	2	<0.01
Ganio (2001) [98]	16	15.5	Williams	4.1	1.25	0.01
Leroi (2001) [99]	6	6	Urgency episodes/1 week	4.8	2.3	>0.05
			FI episodes/1 week	3.2	0.05	>0.05
Matzel (2001) [100]	6	5–66	Wexner	17	2	NR
Rosen (2001) [101]	16	15	FI episodes/3 weeks	6	2	NR
Kenefick (2002) [102]	15	24	FI episodes/1 week	11	0	<0.001
Jarrett (2004) [103]	46	12	FI episodes/1 week	7.5	1	<0.001
Matzel (2004) [104]	34	24	FI episodes/1 week	16.4	2.0	<0.0001
Rasmussen (2004) [105]	45	6	Wexner	16	6	<0.0001
Uludag (2004) [106]	75	12	FI episodes/1 week	7.5	0.67	<0.01
Hetzer (2007) [107]	37	13	Wexner	16	5	<0.01
Holzer (2007) [108]	29	35	FI episodes/3 weeks	7	2	0.002
Melenhorst (2007) [109]	100	36	FI episodes/1 week	31.3	4.8	<0.0001
Tjandra (2008) [110]	53	12	Wexner	16	1.2	<0.0001
Altomare (2009) [111]	52	60	Wexner	15	5	< 0.001
Boyle (2009) [112]	13	3–6	Wexner	12	9	0.0005
			FI episodes/2 weeks	15	3	0.01
Matzel (2009) [113]	9	117.6	Wexner	17	10	<0.007
Dudding (2010) [114]	9	46	FI episodes/1 week	9.9	1.0	0.031
Michelsen (2010) [115]	177	24	Wexner	16	10	<0.0001
Vallet (2010) [116]	23	44	Wexner	16	6.9	NR
Wexner (2010) [90]	120	28	FI episodes/1 week	9.4	2.7	<0.0001
Lim (2011) [117]	41	51	Wexner	11.5	8.0	<0.001
George (2012) [118]	25	114	FI episodes/week	22	0	0.001

NR not reported

prospective, nonrandomized trial in 16 centres in North America and Australia, which was submitted to the FDA for approval of SNS in the United States, 285 patients were evaluated for potential enrollment with stringent guidelines [91]. Exclusion criteria included congenital anorectal malformations, previous rectal surgery (rectopexy, rectal resection, sphincteroplasty within 24 months), external anal sphincter defects greater than 60°, chronic inflammatory bowel disease, visible sequelae of radiation, active anal abscesses or fistulae, and neurologic diseases. Of the 285 patients assessed, 133 patients were candidates for test stimulation. Ninety percent of patients who underwent test stimulation had a successful response (≥50 % improvement) and subsequently underwent permanent implantation. At 1 year of follow-up, 83 % of patients had a greater than 50 % improvement in number of weekly incontinent episodes ($p<0.001$). Perfect continence was achieved in 40 % of patients, and an additional 30 % reported greater than 75 % improvement. These results remained consistent through 3 years of follow-up [120]. The number of weekly incontinent days and number of urgent incontinent episodes per week followed a similar pattern of improvement. In addition, scores on the FISI and on all four domains of the FIQL scale, as well as patients' use of pads, demonstrated statistically and clinically relevant improvements.

To confirm that the clinical benefit derived from SNS was not due to a placebo effect, Leroi et al. conducted a double-blinded crossover study with 27 patients who underwent permanent SNS for faecal incontinence [121]. After implantation, patients were randomized in a double-blind

crossover design to stimulation ON or OFF for 1 month periods. While still blinded, the patients chose the period of stimulation (ON or OFF) that they preferred; the stimulation corresponding to the selected period was continued for 3 months. These investigators found that the number of incontinent episodes was significantly reduced during the ON versus OFF period and the patients had a significantly greater preference for the ON versus OFF period. In the final period of the study, the number of incontinent episodes, Wexner score, ability to postpone defecation, and quality of life improved significantly in patients with the stimulator ON [121].

A recent meta-analysis comparing SNS with maximal conservative therapy included 34 studies and 790 patients, 665 (84.2 %) of whom received a permanent implant [122]. All studies reported a decrease in incontinent episodes per week, with an overall mean difference before and after treatment of −6.83 incontinent episodes (95 % CI −8.05, −5.60; $p<0.001$) with follow-up ranging from 2 to 36 weeks. In the 14 studies reporting pre- and postoperative Wexner scores, a mean difference of −10.57 (95 % CI −11.89, −9.24; $p<0.001$) was observed. All SF-36 outcomes favoured an improved quality of life after SNS implantation except bodily pain, which was not impacted by SNS; and scores on all four domains of the FIQL (lifestyle, coping/behaviour, depression/self-perception, embarrassment) also significantly improved.

Long-Term Outcomes

Several recent reports have demonstrated that the improvement of symptoms and quality of life with SNS is maintained over time. Altomare et al. reported outcomes of 52 patients with more than 5 years follow-up (median, >6 years) and found that nearly 75 % of patients maintained at least a 50 % improvement [112]. Another study reported a sustained effect over a 6-year follow-up period in 10 patients who underwent permanent SNS implantation with a median Wexner score of 7 (range, 2–11) from 20 (range, 12–20) ($p<0.0001$) [116]. George et al. reported their long-term experience with SNS at St. Mark's Hospital [119]. Of 23 patients followed for a median of 9.5 months, full continence was maintained in 12 (48 %) patients; 3 (13 %) patients underwent explantation for loss of efficacy at 48–60 months after permanent implantation; and 3 (13 %) patients died because of unrelated comorbidities. In addition, 9 patients required a device battery change at a mean of 7.25 years.

SNS for Fecal Incontinence with an Associated Sphincter Defect

Considering the favourable long-term outcomes of SNS, for patients with faecal incontinence associated with a sphincter defect, there is a compelling argument for SNS to be the initial treatment instead of an overlapping sphincter repair [123]. Several reports including patients with varying degrees of internal and/or external anal sphincter defects have demonstrated successful outcomes of permanent SNS in this group, indicating that an intact anal sphincter is not a prerequisite for success with SNS. In the recent North American trial, patients with an internal anal sphincter defect had a 65 % success rate compared with 87 % among patients with an intact sphincter [91]. Nonetheless, this success was maintained over a 3-year follow-up. A recent meta-analysis reported five studies that included more than 75 % of patients with sphincter defects and 18 studies that included only patients with an intact sphincter muscle [122]. The number of incontinent episodes per week and Wexner scores improved significantly more in the sphincter intact versus sphincter defect group, whereas the ability to defer defecation was greater in patients with sphincter defects. In a series of 13 patients with sphincter defects who underwent permanent SNS, Boyle et al. observed that SNS results in positive outcomes irrespective of the degree of sphincter disruption [113]. Thus, SNS should certainly not be denied on the basis of a sphincter defect, and it can be considered as the initial treatment for appropriate SNS candidates.

Complications

Compared with the high rates of revision and device removal required for the artificial bowel sphincter and stimulated graciloplasty, the

complications following SNS are uncommon and less severe. Furthermore, even if surgical revision is required, it is a less involved procedure. In a prospective single-centre study including 87 consecutive patients who underwent permanent stimulator implantation, 36 (24.1 %) patients required surgical revision over a mean follow-up of 48.5 months [124]. Several other studies of long-term SNS outcomes report surgical revision rates ranging from 10 to 25 % [91, 93]. A recent meta-analysis reported 6 % pain or local discomfort, 3 % lead displacement or breakage, 3 % infection, and 3 % seroma formation in patients with a permanent SNS implant [90].

A report of infectious complications in the multicenter SNS trial in North America and Australia demonstrated that early infections were less severe and easier to resolve than late infections [125]. This study included 120 patients with a mean follow-up of 28 months. Thirteen (10.8 %) patients had infections, 9 had early infections (within 3 weeks of permanent implantation), and 4 had late infections (13–41 months after implantation). Most of the early infections (78 %) were treated with antibiotics without device explantation. However, all late infections necessitated device explantation, despite administration of intravenous antibiotics. Age, body mass index, and length of operative time for the temporary or permanent implantation were not significantly associated with infection. In a single-centre prospective study, Faucheron et al. reported infection in 4 (4.5 %) patients, necessitating explantation at 2–9 months post-implantation [124]. One infection started with the temporary lead site, whereas the remainder occurred only after permanent SNS implantation. All patients underwent reimplantation of an SNS device at 5–9 months following explantation.

Pain unresponsive to medical management is an uncommon complication (less than 10 %) [124]. If the pain cannot be improved by stimulator setting adjustments, surgical revision with placement of the stimulator in a deeper alternate position may be required. Electrode complications present with sudden worsening of incontinence that does not improve with stimulator setting adjustments and can be confirmed by x-ray. Electrode fracture is suspected when there is excessive impedance in the system (>4,000 Ω). Electrode displacements or fractures are infrequent [124] and require revisional surgery.

However, increased impedance and worsening of faecal incontinence have been reported in patients without electrode breakage. In Faucheron et al.'s series, 4 (5 %) patients presented in this manner, with x-rays confirming correct position and integrity of the electrodes [124]. Re-exploration confirmed an intact circuit, and implantation of a new electrode and new extension cable, attached to the previously implanted stimulator, yielded good results. The authors postulated that the increase in impedance causing failure of the device was secondary to fibrosis surrounding the tip of the electrode, as happens with the dynamic graciloplasty electrode.

Finally, changes in efficacy are addressed by stimulator reprogramming. Govaert et al. retrospectively reviewed their experience with 155 patients with permanent implants and found that 75 % of patients required reprogramming at least once during follow-up (median 28 months) [126]. The mean voltage was significantly increased at 3 months compared with 1 month of follow-up (2.0 V vs 1.8 V, $p<0.001$); however, after 3 months there were no subsequent significant increases in voltage. Fifty-one (33 %) patients required reprogramming at 1–25 % of follow-up visits, 42 (27 %) patients at 26–50 % of follow-up visits, 14 (9 %) patients at 51–75 %, and 9 (6 %) patients at 76–100 % of follow-up visits. Electrode polarity was the most frequently adjusted parameter. This study highlights the importance of close outpatient follow-up with trained clinicians for assessment and reprogramming of SNS in order to achieve optimal outcomes.

SNS is an effective treatment for faecal incontinence that has demonstrable benefits for symptom severity and quality of life outcomes. Technical problems are infrequent and can be easily managed, even when a complete substitution of the device is required. Furthermore, as this is a minimally invasive procedure, it is suitable for many patients, including those affected by severe comorbidities that would preclude other more invasive surgery.

Stimulation: Posterior Tibial Nerve Stimulation

As with SNS, posterior tibial nerve stimulation (PTNS) was first described for urinary incontinence [127]. The tibial nerve is a mixed nerve composed of L4–S3 fibres, and it originates from the same spinal segments that innervate the pelvic floor [92].

Percutaneous Technique

A fine gauge needle is percutaneously inserted, just above and medial to the ankle, next to the posterior tibial nerve, and a surface electrode is placed near the arch of the foot. The needle and electrode are connected to a low-voltage stimulator. Stimulation of the posterior tibial nerve produces a motor (plantar flexion or fanning of the toes) and/or sensory (tingling in the ankle, foot, or toes) response. Initial treatment usually consists of 12 outpatient sessions lasting 30 min each, typically 1 week apart. Treatment may be repeated if required [128].

Outcomes

In 2003, Shafik et al. first reported PTNS for the treatment of faecal incontinence [129]. Vitton et al. reported significant benefit in a cohort of 24 patients, 46 % of whom showed sustained improvement by 1 year after completion of stimulation [130]. Hotouras et al. reported the largest study to date, which included 100 patients evaluated prospectively [131]. They found that Wexner scores and quality of life were significantly improved following 12 sessions of PTNS. Although long-term results are lacking, these short-term results are promising. PTNS can be performed via percutaneous and transcutaneous routes; however, the optimal route is still debated. George et al. compared the efficacy of both approaches to sham stimulation in a randomized trial including 30 patients [132]. These investigators found that 82 % of patients who underwent percutaneous PTNS had a 50 % or greater reduction in weekly incontinent episodes, compared with 45 % of patients who underwent transcutaneous PTNS and 12.5 % of patients who underwent sham stimulation.

Conclusion

From simple, office-based procedures with low durability to sophisticated, technically demanding, and highly complicated surgical procedures, the ideal treatment for faecal incontinence requires an individualized approach. The introduction of new technology is encouraging, and it is hoped that it will advance these much needed procedures. Despite this plethora of exciting advances, a stoma still remains the best option in many patients with end-stage faecal incontinence.

References

1. Lewis SJ, Heaton KW. Stool form scale as a useful guide to intestinal transit time. Scand J Gastroenterol. 1997;32(9):920–4.
2. Jorge JMN, Wexner SD. Etiology and management of faecal incontinence. Dis Colon Rectum 1993;36:77–97.
3. Goode PS, Burgio KL, Halli AD, et al. Prevalence and correlates of faecal incontinence in community-dwelling older adults. J Am Geriatr Soc. 2005;53:629–35.
4. Bliss DZ, Jung HJ, Savok K, et al. Supplementation with dietary fiber improves faecal incontinence. Nurs Res. 2001;50:203–13.
5. Ehrenpreis ED, Chang D, Eichenwald E. Pharmacotherapy for faecal incontinence: a review. Dis Colon Rectum. 2007;50(5):641–9.
6. Palmer KR, Corbett CL, Holdsworth CD. Double-blind crossover study comparing loperamide, codeine and diphenoxylate in the treatment of chronic diarrhea. Gastroenterology. 1980;79:1272–5.
7. Oberwalder M, Connor J, Wexner SD. Meta-analysis to determine the incidence of obstetric anal sphincter damage. Br J Surg. 2003;90:1333–7.
8. Gilliland R, Altomare DF, Moreira Jr H, et al. Pudendal neuropathy is predictive of failure following anterior overlapping sphincteroplasty. Dis Colon Rectum. 1998;41(12):1516–22.
9. Goetz LH, Lowry AC. Overlapping sphincteroplasty: is it the standard of care? Clin Colon Rectal Surg. 2005;18(1):22–31.
10. Zetterström J, López A, Anzén B, et al. Anal sphincter tears at vaginal delivery: risk factors and clinical outcome of primary repair. Obstet Gynecol. 1999;94(1):21–8.

11. Hasegawa H, Yoshioka K, Keighley MR. Randomized trial of faecal diversion for sphincter repair. Dis Colon Rectum. 2000;43(7):961–4.
12. Mahony R, Behan M, O'Herlihy C, O'Connell PR. Randomized, clinical trial of bowel confinement vs. laxative use after primary repair of a third-degree obstetric anal sphincter tear. Dis Colon Rectum. 2004;47(1):12–7.
13. Fleshman JW, Peters WR, Shemesh EI, et al. Anal sphincter reconstruction: anterior overlapping muscle repair. Dis Colon Rectum. 1991;34(9):739–43.
14. Wexner SD, Marchetti F, Jagelman DG. The role of sphincteroplasty for faecal incontinence reevaluated: a prospective physiologic and functional review. Dis Colon Rectum. 1991;34(1):22–30.
15. Engel AF, Kamm MA, Sultan AH, et al. Anterior anal sphincter repair in patients with obstetric trauma. Br J Surg. 1994;81(8):1231–4.
16. Halverson AL, Hull TL. Long-term outcome of overlapping anal sphincter repair. Dis Colon Rectum. 2002;45(3):345–8.
17. Zutshi M, Tracey TH, Bast J, et al. Ten-year outcome after anal sphincter repair for faecal incontinence. Dis Colon Rectum. 2009;52(6):1089–94.
18. Gordon PH, Nivatvongs S. Principles and practice of surgery for the colon, rectum, and anus. 3rd ed. London: Informa Healthcare; 2007.
19. Matsuoka H, Mavrantonis C, Wexner SD, Oliveira L, Gilliland R, Pikarsky A. Postanal repair for faecal incontinence – is it worthwhile? Dis Colon Rectum. 2000;43(11):1561–7.
20. Mackey P, Mackey L, Kennedy ML, et al. Postanal repair – do the long-term results justify the procedure? Colorectal Dis. 2010;12(4):367–72.
21. Takahashi T, Garcia-Osogobio S, Valdovinos MA, et al. Radio-frequency energy delivery to the anal canal for the treatment of faecal incontinence. Dis Colon Rectum. 2002;45(7):915–22.
22. Efron JE, Corman ML, Fleshman J, et al. Safety and effectiveness of temperature-controlled radio-frequency energy delivery to the anal canal (Secca procedure) for the treatment of faecal incontinence. Dis Colon Rectum. 2003;46(12):1606–16.
23. Takahashi T, Garcia-Osogobio S, Valdovinos MA, et al. Extended two-year results of radio-frequency energy delivery for the treatment of faecal incontinence (the Secca procedure). Dis Colon Rectum. 2003;46(6):711–5.
24. Takahashi-Monroy T, Morales M, Garcia-Osogobio S, et al. SECCA procedure for the treatment of faecal incontinence: results of five-year follow-up. Dis Colon Rectum. 2008;51(3):355–9.
25. Lefebure B, Tuech JJ, Bridoux V, et al. Temperature-controlled radio frequency energy delivery (Secca procedure) for the treatment of faecal incontinence: results of a prospective study. Int J Colorectal Dis. 2008;23(10):993–7.
26. Ruiz D, Pinto RA, Hull TL, et al. Does the radiofrequency procedure for faecal incontinence improve quality of life and incontinence at 1-year follow-up? Dis Colon Rectum. 2010;53(7):1041–6.
27. Shafik A. Polytetrafluoroethylene injection for treatment of partial faecal incontinence. Int Surg. 1993;78(2):159–61.
28. Shafik A. Perianal injection of autologous fat for treatment of sphincteric incontinence. Dis Colon Rectum. 1995;38(6):583–7.
29. Malouf AJ, Vaizey CJ, Norton CS, Kamm MA. Internal anal sphincter augmentation for faecal incontinence using injectable silicone biomaterial. Dis Colon Rectum. 2001;44(4):595–600.
30. Davis K, Kumar D, Poloniecki J. Preliminary evaluation of an injectable anal sphincter bulking agent (Durasphere) in the management of faecal incontinence. Aliment Pharmacol Ther. 2003;18(2):237–43.
31. Tjandra JJ, Lim JF, Hiscock R, Rajendra P. Injectable silicone biomaterial for faecal incontinence caused by internal anal sphincter dysfunction is effective. Dis Colon Rectum. 2004;47(12):2138–46.
32. Chan MK, Tjandra JJ. Injectable silicone biomaterial (PTQ) to treat faecal incontinence after hemorrhoidectomy. Dis Colon Rectum. 2006;49(4):433–9.
33. Stojkovic SG, Lim M, Burke D, Finan PJ, Sagar PM. Intra-anal collagen injection for the treatment of faecal incontinence. Br J Surg. 2006;93(12):1514–8.
34. de la Portilla F, Fernández A, et al. Evaluation of the use of PTQ implants for the treatment of incontinent patients due to internal anal sphincter dysfunction. Colorectal Dis. 2008;10(1):89–94.
35. Maeda Y, Vaizey CJ, Kamm MA. Pilot study of two new injectable bulking agents for the treatment of faecal incontinence. Colorectal Dis. 2008;10(3):268–72.
36. Soerensen MM, Lundby L, Buntzen S, Laurberg S. Intersphincteric injected silicone biomaterial implants: a treatment for faecal incontinence. Colorectal Dis. 2009;11(1):73–6.
37. Tjandra JJ, Chan MK, Yeh HC. Injectable silicone biomaterial (PTQ) is more effective than carbon-coated beads (Durasphere) in treating passive faecal incontinence – a randomized trial. Colorectal Dis. 2009;11(4):382–9.
38. Graf W, Mellgren A, Matzel KE, et al. Efficacy of dextranomer in stabilised hyaluronic acid for treatment of faecal incontinence: a randomised, sham-controlled trial. Lancet. 2011;377(9770):997–1003.
39. Maeda Y, Laurberg S, Norton C. Perianal injectable bulking agents as treatment for faecal incontinence in adults. Cochrane Database Syst Rev. 2010;(5):CD007959.
40. Chetwood CH. Plastic operation of the sphincter ani with report of a case. Med Rec. 1902;61:529.
41. Cera S, Wexner SD. Muscle transposition: does it still have a role? Clin Colon Rectal Surg. 2005;18(1):46–54.
42. Devesa JM, Madrid JM, Gallego BR, Vicente E, Nuño J, Enríquez JM. Bilateral gluteoplasty for faecal incontinence. Dis Colon Rectum. 1997;40(8):883–8.
43. Pickrell KI, Broadbent TR, Masters FW, et al. Construction of rectal sphincter and restoration of

anal continence by transplanting the gracilis muscle. Report of 4 cases in children. Ann Surg. 1952;135:853–62.
44. Corman ML. Gracilis muscle transposition for anal incontinence: late results. Br J Surg. 1985;72:21.
45. Christiansen J, Sørensen M, Rasmussen OO. Gracilis muscle transposition for faecal incontinence. Br J Surg. 1990;77(9):1039–40.
46. Kumar D, Hutchinson R, Grant E. Bilateral gracilis neosphincter construction for treatment of faecal incontinence. Br J Surg. 1995;82:1645.
47. Salmons S. The adaptive response of skeletal muscle to increased use. Muscle Nerve. 1981;4:94–110.
48. Baeten C, Spaans F, Fluks A. An implanted neuromuscular stimulator for faecal continence. Dis Colon Rectum. 1988;31:134–7.
49. Person B, Wexner SD. Advances in the surgical treatment of faecal incontinence. Surg Innov. 2005;12:7–21.
50. Wexner SD, Gonzalez-Padron A, Rius J, et al. Stimulated gracilis neosphincter operation. Initial experience, pitfalls, and complications. Dis Colon Rectum. 1996;39(9):957–64.
51. Baeten CG, Bailey HR, Bakka A, et al. Safety and efficacy of dynamic graciloplasty for faecal incontinence: report of a prospective, multicenter trial. Dynamic Graciloplasty Therapy Study Group. Dis Colon Rectum. 2000;43(6):743–51.
52. Wexner SD, Baeten C, Bailey R, et al. Long-term efficacy of dynamic graciloplasty for faecal incontinence. Dis Colon Rectum. 2002;45(6):809–18.
53. Chapman AE, Geerdes B, Hewett P, et al. Systematic review of dynamic graciloplasty in the treatment of faecal incontinence. Br J Surg. 2002;89(2):138–53.
54. Christiansen J, Rasmussen OO, Lindorff-Larsen K. Dynamic graciloplasty for severe anal incontinence. Br J Surg. 1998;85(1):88–91.
55. Sielezneff I, Malouf AJ, Bartolo DC, Pryde A, Douglas S. Dynamic graciloplasty in the treatment of patients with faecal incontinence. Br J Surg. 1999;86(1):61–5.
56. Mavrantonis C, Billotti VL, Wexner SD. Stimulated graciloplasty for faecal incontinence: critical influence of the method of stimulation. Dis Colon Rectum. 1999;42(4):497–504.
57. Mander BJ, Wexner SD, Williams NS, et al. Preliminary results of a multicenter trial of the electrically stimulated gracilis neoanal sphincter. Br J Surg. 1999;86(12):1543–9.
58. Madoff RD, Rosen HR, Baeten CG, et al. Safety and efficacy of dynamic muscle plasty for anal incontinence: lessons from a prospective, multicenter trial. Gastroenterology. 1999;116(3):549–56.
59. Konsten J, Rongen MJ, Ogunbiyi OA, et al. Comparison of epineural and intramuscular nerve electrodes for stimulated graciloplasty. Dis Colon Rectum. 2001;44(4):581–6.
60. Bresler L, Reibel N, Brunaud L, Sielezneff I, Rouanet P, Rullier E, Slim K. Dynamic graciloplasty in the treatment of severe faecal incontinence. French multicentric retrospective study. Ann Chir. 2002;127(7):520–6.
61. Rongen MJ, Uludag O, El Naggar K, et al. Long-term follow-up of dynamic graciloplasty for faecal incontinence. Dis Colon Rectum. 2003;46(6):716–21.
62. Penninckx F, Belgian Section of Colorectal Surgery. Belgian experience with dynamic graciloplasty for faecal incontinence. Br J Surg. 2004;91(7):872–8.
63. Matzel KE, Madoff RD, LaFontaine LJ, et al. Dynamic Graciloplasty Therapy Study Group. Complications of dynamic graciloplasty: incidence, management, and impact on outcome. Dis Colon Rectum. 2001;44(10):1427–35.
64. Person B, Kaidar-Person O, Wexner SD. Novel approaches in the treatment of faecal incontinence. Surg Clin North Am. 2006;86:969–86.
65. Lehur PA, Meurette G, Torre F. The artificial bowel sphincter in the treatment of severe faecal incontinence in adults. In: Ratto C et al., editors. Fecal incontinence. Milan: Springer; 2007. p. 193–203.
66. Wong WD, Jensen LL, Bartolo DCC, Rothenberger DA. Artificial anal sphincter. Dis Colon Rectum. 1996;39:1345–51.
67. Lehur PA, Glemain P, Bruley des Varannes S, Buzelin JM, Leborgne J. Outcome of patients with an implanted artificial anal sphincter for severe faecal incontinence. A single institution report. Int J Colorectal Dis. 1998;13:88–92.
68. Vaizey CJ, Kamm MA, Gold DM, et al. Clinical, physiological, and radiological study of a new purpose-designed artificial bowel sphincter. Lancet. 1998;352:105–9.
69. Christiansen J, Rasmussen OO, Lindorff-Larsen K. Long-term results of artificial anal sphincter implantation for severe anal incontinence. Ann Surg. 1999;230:45–8.
70. Lehur PA, Roig JV, Duinslaeger M. Artificial anal sphincter: prospective clinical and manometric evaluation. Dis Colon Rectum. 2000;43:1100–6.
71. O'Brien PE, Skinner S. Restoring control: the Acticon Neosphincter artificial bowel sphincter in the treatment of anal incontinence. Dis Colon Rectum. 2000;43:1213–6.
72. Altomare DF, Dodi G, La Torre F, et al. Multicentre retrospective analysis of the outcome of artificial anal sphincter implantation for severe faecal incontinence. Br J Surg. 2001;88:1481–6.
73. Lehur PA, Zerbib F, Neunlist M, Glemain P, Bruley des Varannes S. Comparison of quality of life and anorectal function after artificial sphincter implantation. Dis Colon Rectum. 2002;45:508–13.
74. Devesa JM, Rey A, Hervas PL, et al. Artificial anal sphincter: complications and functional results of a large personal series. Dis Colon Rectum. 2002;45(9):1154–63.
75. Ortiz H, Armendariz P, DeMiguel M, Ruiz MD, Alos R, Roig JV. Complications and functional outcome following artificial anal sphincter implantation. Br J Surg. 2002;89(7):877–81.

76. Wong WD, Congliosi SM, Spencer MP, et al. The safety and efficacy of the artificial bowel sphincter for faecal incontinence: results from a multicenter cohort study. Dis Colon Rectum. 2002;45(9):1139–53.
77. Michot F, Costaglioli B, Leroi AM, Denis P. Artificial anal sphincter in severe faecal incontinence: outcome of prospective experience with 37 patients in one institution. Ann Surg. 2003;237:52–6.
78. Parker SC, Spencer MP, Madoff RD, Jensen LL, Wong WD, Rothenberger DA. Artificial bowel sphincter: long-term experience at a single institution. Dis Colon Rectum. 2003;46(6):722–9.
79. Casal E, San Ildefonsol A, Carracedo R, Facal C, Sánchez JA. Artificial bowel sphincter in severe anal incontinence. Colorectal Dis. 2004;6:180–4.
80. Ruiz-Carmona MD, Company RA, Roig Vila JV, et al. Long-term results of artificial bowel sphincter for the treatment of severe faecal incontinence. Are they what we hoped for? Colorectal Dis. 2008;11:831–7.
81. Wexner SD, Jin HY, Weiss EG, Nogueras JJ, Li VK. Factors associated with failure of the artificial bowel sphincter: a study of over 50 cases from Cleveland Clinic Florida. Dis Colon Rectum. 2009;52(9):1550–7.
82. Christiansen J, Lorentzen M. Implantation of artificial sphincter for anal incontinence. Lancet. 1987;2:244–5.
83. Mundy L, Merlin TL, Maddern GJ, Hiller JE. Systematic review of safety and effectiveness of an artificial bowel sphincter for faecal incontinence. Br J Surg. 2004;91:665–72.
84. Belyaev O, Muller C, Uhl W. Neosphincter surgery for faecal incontinence: a critical and unbiased review of the relevant literature. Surg Today. 2006;36(4):295–303.
85. Winslett MC, Allan A, Ambrose NS. Anorectal sepsis as a presentation of occult rectal and systemic disease. Dis Colon Rectum. 1988;31:597–600.
86. O'Brien PE, Dixon JB, Skinner S, Laurie C, Khera A, Fonda D. A prospective, randomized, controlled clinical trial of placement of the artificial bowel sphincter (Acticon Neosphincter) for the control of faecal incontinence. Dis Colon Rectum. 2004;47(11):1852–60.
87. Tejirian T, Kaminski A, Abbas MA. Intra-abdominal erosion of artificial bowel sphincter reservoir. Int J Colorectal Dis. 2007;22(7):849–50.
88. Matzel KE, Stadelmaier U, Hohenfellner M, Gall FP. Electrical stimulation of sacral spinal nerves for treatment of faecal incontinence. Lancet. 1995;346:1124–7.
89. Dudding TC, Parés D, Vaizey CJ, Kamm MA. Comparison of clinical outcome between open and percutaneous lead insertion for permanent sacral nerve neurostimulation for the treatment of faecal incontinence. Dis Colon Rectum. 2009;52:463–8.
90. Hetzer FH. Fifteen years of sacral nerve stimulation: from an open procedure to a minimally invasive technique. Colorectal Dis. 2011;13 Suppl 2:1–4.
91. Wexner SD, Coller JA, Devroede G, et al. Sacral nerve stimulation for faecal incontinence: results of a 120-patient prospective multicenter study. Ann Surg. 2010;251(3):441–9.
92. O'Connell PR. The physiology of sacral neuromodulation. Paper presented at the 23rd annual international colorectal disease symposium, Fort Lauderdale, 14–18 Feb, 2012.
93. Griffin KM, Pickering M, O'Herlihy C, O'Connell PR, Jones JF. Sacral nerve stimulation increases activation of the primary somatosensory cortex by anal canal stimulation in an experimental model. Br J Surg. 2011;98(8):1160–9.
94. Sheldon R, Kiff ES, Clarke A, Harris ML, Hamdy S. Sacral nerve stimulation reduces corticoanal excitability in patients with faecal incontinence. Br J Surg. 2005;92:1423–31.
95. Gooneratne ML, Facer P, Knowles CH, et al. Normalization of substance P levels in rectal mucosa of patients with faecal incontinence treated successfully by sacral nerve stimulation. Br J Surg. 2008;95:477–83.
96. Abdel-Halim MR, Crosbie J, Engledow A, Windsor A, Cohen CR, Emmanuel AV. Temporary sacral nerve stimulation alters rectal sensory function: a physiological study. Dis Colon Rectum. 2011;54(9):1134–40.
97. Otto SD, Burmeister S, Buhr HJ, Kroesen A. Sacral nerve stimulation induces changes in the pelvic floor and rectum that improve continence and quality of life. J Gastrointest Surg. 2010;14(4):636–44.
98. Malouf AJ, Vaizey CJ, Nicholls RJ, Kamm MA. Permanent sacral nerve stimulation for faecal incontinence. Ann Surg. 2000;232(1):143–8.
99. Ganio E, Ratto C, Masin A, et al. Neuromodulation for faecal incontinence: outcome in 16 patients with definitive implant. The initial Italian Sacral Neurostimulation Group (GINS) experience. Dis Colon Rectum. 2001;44(7):965–70.
100. Leroi AM, Michot F, Grise P, Denis P. Effect of sacral nerve stimulation in patients with faecal and urinary incontinence. Dis Colon Rectum. 2001;44(6):779–89.
101. Matzel KE, Stadelmaier U, Hohenfellner M, Hohenberger W. Chronic sacral spinal nerve stimulation for faecal incontinence: long-term results with foramen and cuff electrodes. Dis Colon Rectum. 2001;44:59–66.
102. Rosen HR, Urbarz C, Holzer B, Novi G, Schiessel R. Sacral nerve stimulation as a treatment for faecal incontinence. Gastroenterology. 2001;121:536–41.
103. Kenefick NJ, Vaizey CJ, Cohen RC, Nicholls RJ, Kamm MA. Medium-term results of permanent sacral nerve stimulation for faecal incontinence. Br J Surg. 2002;89:896–901.
104. Jarrett ME, Varma JS, Duthie GS, Nicholls RJ, Kamm MA. Sacral nerve stimulation for faecal incontinence in the UK. Br J Surg. 2004;91(6):755–61.
105. Matzel KE, Kamm MA, Stösser M, et al. Sacral spinal nerve stimulation for faecal incontinence: multicentre study. Lancet. 2004;363(9417):1270–6.

106. Rasmussen OO, Buntzen S, Sørensen M, Laurberg S, Christiansen J. Sacral nerve stimulation in faecal incontinence. Dis Colon Rectum. 2004;47:1158–63.
107. Uludag O, Koch SM, van Gemert WG, Dejong CH, Baeten CG. Sacral neuromodulation in patients with faecal incontinence: a single-center study. Dis Colon Rectum. 2004;47(8):1350–7.
108. Hetzer FH, Hahnloser D, Clavien PA, Demartines N. Quality of life and morbidity after permanent sacral nerve stimulation for faecal incontinence. Arch Surg. 2007;142(1):8–13.
109. Holzer B, Rosen HR, Novi G, Ausch C, Hölbling N, Schiessel R. Sacral nerve stimulation for neurogenic faecal incontinence. Br J Surg. 2007;94(6):749–53.
110. Melenhorst J, Koch SM, Uludag O, van Gemert WG, Baeten CG. Sacral neuromodulation in patients with faecal incontinence: results of the first 100 permanent implantations. Colorectal Dis. 2007;9(8):725–30.
111. Tjandra JJ, Chan MK, Yeh CH, Murray-Green C. Sacral nerve stimulation is more effective than optimal medical therapy for severe faecal incontinence: a randomized, controlled study. Dis Colon Rectum. 2008;51(5):494–502.
112. Altomare DF, Ratto C, Ganio E, Lolli P, Masin A, Villani RD. Long-term outcome of sacral nerve stimulation for faecal incontinence. Dis Colon Rectum. 2009;52(1):11–7.
113. Boyle DJ, Knowles CH, Lunniss PJ, Scott SM, Williams NS, Gill KA. Efficacy of sacral nerve stimulation for faecal incontinence in patients with anal sphincter defects. Dis Colon Rectum. 2009;52(7):1234–9.
114. Matzel KE, Lux P, Heuer S, Besendörfer M, Zhang W. Sacral nerve stimulation for faecal incontinence: long-term outcome. Colorectal Dis. 2009;11(6):636–41.
115. Dudding TC, Parés D, Vaizey CJ, Kamm MA. Sacral nerve stimulation for the treatment of faecal incontinence related to dysfunction of the internal anal sphincter. Int J Colorectal Dis. 2010;25(5):625–30.
116. Michelsen HB, Thompson-Fawcett M, Lundby L, Krogh K, Laurberg S, Buntzen S. Six years of experience with sacral nerve stimulation for faecal incontinence. Dis Colon Rectum. 2010;53:414–21.
117. Vallet C, Parc Y, Lupinacci R, Shields C, Parc R, Tiret E. Sacral nerve stimulation for faecal incontinence: response rate, satisfaction and the value of preoperative investigation in patient selection. Colorectal Dis. 2010;12:247–53.
118. Lim JT, Hastie IA, Hiscock RJ, Shedda SM. Sacral nerve stimulation for faecal incontinence: long-term outcomes. Dis Colon Rectum. 2011;54(8):969–74.
119. George AT, Kalmar K, Panarese A, Dudding TC, Nicholls RJ, Vaizey CJ. Long-term outcomes of sacral nerve stimulation for faecal incontinence. Dis Colon Rectum. 2012;55(3):302–6.
120. Mellgren A, Wexner SD, Coller JA, et al. Long-term efficacy and safety of sacral nerve stimulation for faecal incontinence. Dis Colon Rectum. 2011;54(9):1065–75.
121. Leroi AM, Parc Y, Lehur PA, et al. Efficacy of sacral nerve stimulation for faecal incontinence: results of a multicenter double-blind crossover study. Ann Surg. 2005;242(5):662–9.
122. Tan E, Ngo NT, Darzi A, Shenouda M, Tekkis PP. Meta-analysis: sacral nerve stimulation versus conservative therapy in the treatment of faecal incontinence. Int J Colorectal Dis. 2011;26(3):275–94.
123. Hull TL. Sacral neuromodulation stimulation in faecal incontinence. Int Urogynecol J. 2010;21(12):1565–8.
124. Faucheron JL, Voirin D, Badic B. Sacral nerve stimulation for faecal incontinence: causes of surgical revision from a series of 87 consecutive patients operated on in a single institution. Dis Colon Rectum. 2010;53(11):1501–7.
125. Wexner SD, Hull T, Edden Y, et al. Infection rates in a large investigational trial of sacral nerve stimulation for faecal incontinence. J Gastrointest Surg. 2010;14(7):1081–9.
126. Govaert B, Rietveld MP, van Gemert WG, Baeten CG. The role of reprogramming in sacral nerve modulation for faecal incontinence. Colorectal Dis. 2011;13(1):78–81.
127. McGuire EJ, Zhang SC, Horwinski ER, Lytton B. Treatment of motor and sensory detrusor instability by electrical stimulation. J Urol. 1983;129(1):78–9.
128. National Institute for Health and Clinical Excellence. Percutaneous tibial nerve stimulation for faecal incontinence. http://guidance.nice.org.uk/IPG395. Accessed 25 June 2012.
129. Shafik A, Ahmed I, El-Sibai O, Mostafa RM. Percutaneous peripheral neuromodulation in the treatment of faecal incontinence. Eur Surg Res. 2003;35(2):103–7.
130. Vitton V, Damon H, Roman S, Mion F. Transcutaneous electrical posterior tibial nerve stimulation for faecal incontinence: effects on symptoms and quality of life. Int J Colorectal Dis. 2010;25(8):1017–20.
131. Hotouras A, Thaha MA, Boyle D, et al. Short-term outcome following percutaneous tibial nerve stimulation (PTNS) for faecal incontinence: a single-centre prospective study. Colorectal Dis. 2012;14(9):1101–5. doi:10.1111/j.1463-1318.2011.02906.x.
132. George A, Kalmar K, Kokopoulis K, et al. A prospective single blinded placebo controlled study into the role of percutaneous and transcutaneous tibial nerve stimulation for faecal incontinence. Gut. 2011;60:A157–8.

Haemorrhoids: Anatomy, Pathophysiology and Presentation

13

Ayesha Noorani and Emin Carapeti

Introduction

The word haemorrhoid is derived from the Greek word *haem* (blood) and *rhoos* (flow). The term piles is derived from Latin *pila*, meaning swelling or ball.

Historical references to haemorrhoids exist as early as the Babylonian and Egyptian eras. Haemorrhoids have long since troubled mankind and have even been known to change the course of history. It is rumoured that Napoleon's haemorrhoids prevented him from mounting his horse before the battle of Waterloo [1].

Hippocrates wrote about them in detail in the Hippocratic Treatises in 460–375 BC. He believed that the presence of 'bile or phlegm' in the veins of the rectum was responsible for heating the blood and resulting in bleeding and bursting of the haemorrhoid. He described several methods of cautery including the use of red hot iron. He stated that when 'the cautery is applied the patient's head and hands should be held so that he may not stir, but he himself should cry out for this will make the rectum project more'. Luckily advancements in medicine have resulted in kinder therapeutic measures of dealing with haemorrhoids.

The Romans made less innovative contributions to the diagnostic and therapeutic management of haemorrhoids, often using methods employed by the Egyptians and Greeks. Galen, physician of the Roman emperor Marcus Aurelius, described the tying of the haemorrhoid with a thread to shrivel it up. The Arab physician Al-Zahrawy in the Byzantine era described the use of cautery irons and thread ligation of haemorrhoids.

The master surgeons era and the subsequent barber surgeons era are scarce on haemorrhoids. Further advancements were seen in 1835 when Frederick Salmon founded St Mark's hospital in London. Subsequently Milligan and Morgan described the definitive operative management of haemorrhoids [2].

Despite advances in the treatment and management of haemorrhoids, our basic understanding of why they occur remains limited. Several theories exist, but often without scientific evidence. Hence, a thorough understanding of the development and anatomy of the anal canal is essential.

Anatomy

Normal Anatomy and Development

External haemorrhoids are derived from ectoderm and are covered by squamous epithelium. In contrast, internal haemorrhoids are derived from embryonic endoderm and lined with columnar epithelium of anal mucosa.

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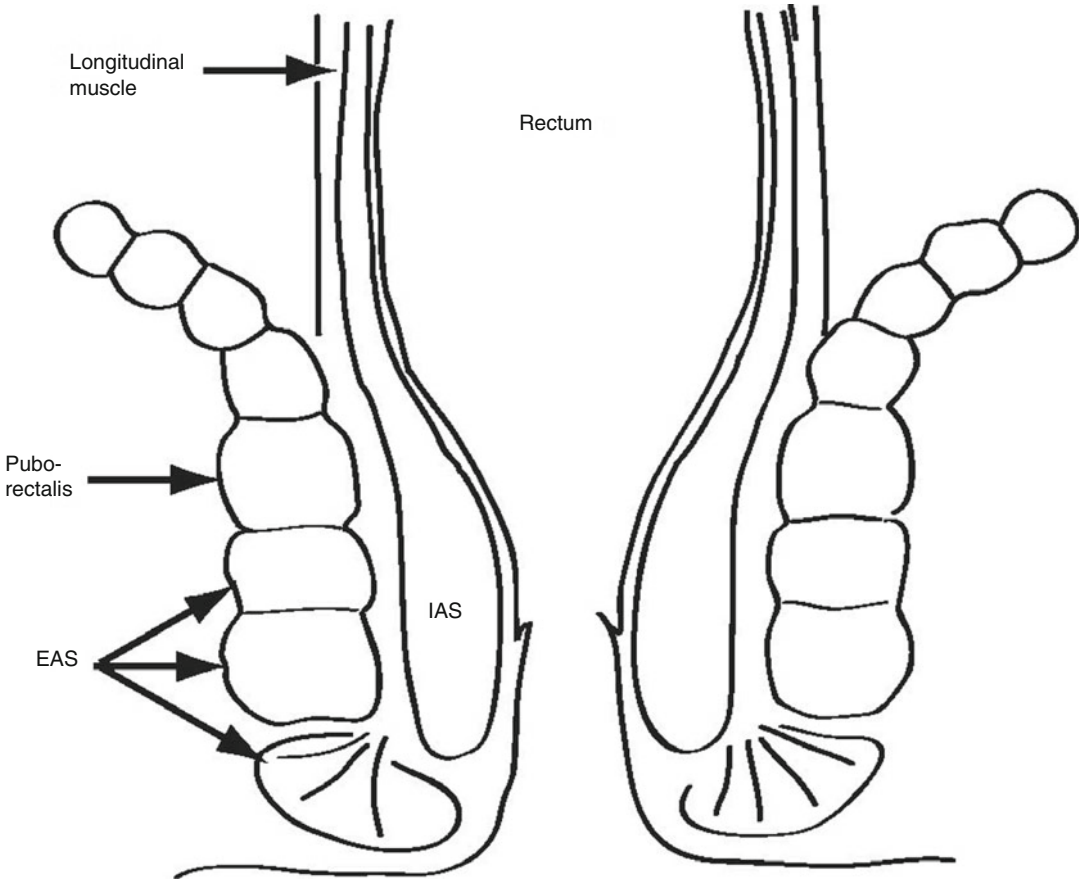

Fig. 13.1 Sagittal section of the anatomy of the anal canal

The normal anatomy of the anal canal is shown in Fig. 13.1. During foetal development, mucosa and submucosa above the dentate line acquire folds known as anal cushions. There are usually three anal cushions, traditionally at left lateral, right posterior and right anterior positions, or 3, 7 and 11 o'clock positions. Thomson et al. revealed that only 19 % of control subjects had this traditional arrangement [3].

Proximally these extend as primary rectal foldings and are separated and covered by the rectal columns of Morgagni [4]. Caudal to this the anal canal is lined by stratified squamous epithelium. The anal cushions are made up of mucosa, submucosa, fibroelastic connective tissue, smooth muscle and blood vessels.

Function of Anal Cushions

The function of the anal cushions has been the topic of much interest and debate. The fact that they can vary their size, given their vascular components, suggests that they have a role in continence. Hence, if we propose that anal cushions are physiological, then only when they become pathological should they be referred to as haemorrhoids.

Studies suggest that the internal sphincter alone cannot completely close the anal canal even during periods of maximal contraction. Lestar et al. demonstrated that the anal cushions have to fill up an intrasphincteric gap of at least 7–8 mm in diameter [5]. Vascular filling contributes to

15–20 % of resting anal pressure as evident from electrophysiological studies [6]. Anal cushions probably provide a plug to maintain continence, and indeed haemorrhoidectomy has been shown to impair continence to infused saline [7].

Mechanisms by which anal cushions facilitate defecation have been studied with much interest. Passive pressure from stool may contribute. In addition active mechanisms include (1) anal dilatation which in turn reduces the height of anal cushions and (2) contraction of the subepithelial smooth muscle layers and the conjoint longitudinal layer resulting in displacement of the anal cushions. The conjoint longitudinal layer has both voluntary and involuntary components that function as a complex to control anal action during defecation.

Anatomy of Haemorrhoids

With age, the sphincter complex changes. The smooth muscle layer becomes replaced by connective tissue. In addition the submucosa thickens and the connective issue becomes looser and more fragmented [8]. The sphincter complex looses strength and the anal cushions are less protected from the forces placed upon them. The cushions become more vulnerable to trauma and migrate downwards with the effect of gravity, and the loss of muscular elements of the submucosa may reduce contractility and strength of the walls of the vascular components rendering them more likely to bleed or thrombose. Figure 13.2 illustrates anatomical differences between first-, second- and third-degree haemorrhoids.

Histological analysis of haemorrhoidectomy specimens reveals squamous metaplasia and dense capillary vasculature. There is reduced collagen to protein ratio and a reduced type I to type III collagen ratio. Hence, there may be a disorder of collagen metabolism that needs attention [9].

Blood Supply

Arteries

The arterial supply of the haemorrhoidal plexus is from the superior, middle and inferior haemorrhoidal arteries. The superior haemorrhoidal artery is a continuation of the inferior mesenteric

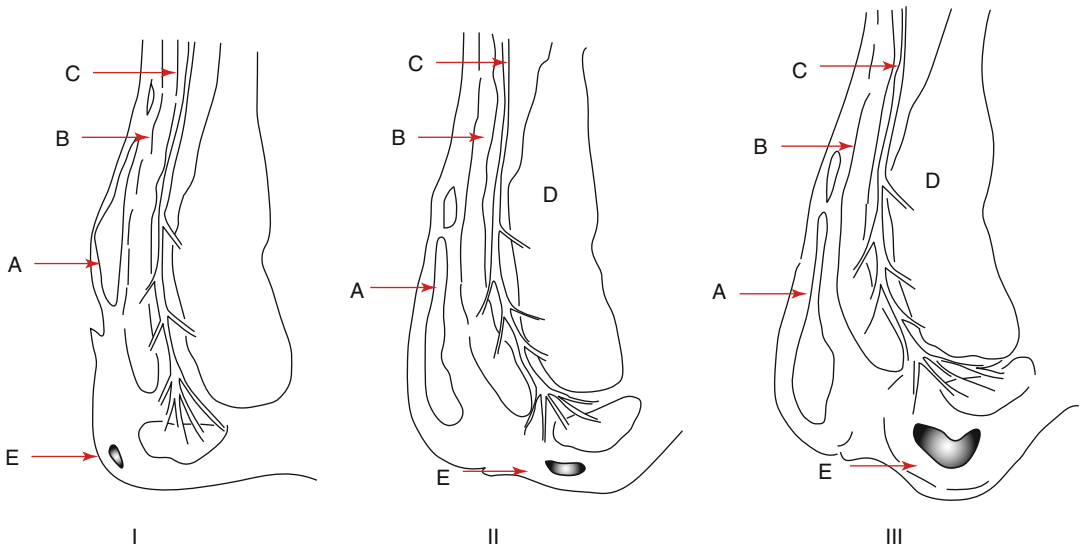

Fig. 13.2 Classification of haemorrhoids: sagittal section of the anal canal demonstrating the descent of the anal cushions. *A* Submucosal plexus, *B* Internal anal sphincter, *C* Longitudinal muscle, *D* External anal sphincter, *E* Anal cushion

artery and descends into the pelvis in the sigmoid mesocolon. It divides into two further branches on either side of the rectum at the level of the third sacral vertebra. Further smaller branches traverse the rectal wall at the submucosal level to supply the internal sphincter and anastomose with branches of the middle and inferior haemorrhoidal arteries. The middle rectal artery arises from the anterior trunk of the internal iliac artery with the inferior vesical artery, whilst the inferior rectal artery arises above the ischial tuberosity from the internal pudendal artery, which is itself a branch of the internal iliac artery.

This rich anastomosis makes rectal ischaemia uncommon and bleeding from haemorrhoids a distinct problem.

Veins

The venous return of the haemorrhoidal plexus follows the arterial pattern. The superior rectal vein drains into the inferior mesenteric vein, which itself drains into the portal vein. The middle and inferior haemorrhoidal veins drain directly to the internal iliac veins and via the pudendal vein to the internal iliac vein, respectively. They form a site of portosystemic venous anastomosis which is relevant in patients with portal hypertension. Haemorrhoids in these patients are far rarer than true rectal varices. This portosystemic anastomosis is also the likely route of rare cases of mesenteric or portal pyaemia, portal thrombosis and sepsis and even liver abscesses seen after injection sclerotherapy, banding or ligation of haemorrhoids [10].

Lymphatic Drainage

The upper half of the rectum has lymphatic drainage which follows the superior haemorrhoidal vessels through pararectal nodes to nodes in the lower sigmoid mesocolon and along the IMA [11].

The lymphatics of the lower half of the rectum and the anal canal proximal to the dentate line accompany the middle rectal vessels to the internal iliac nodes.

Lymphatics of the anal canal below the dentate line drain into superficial inguinal nodes.

Innervation

The inferior rectal nerve (S2, S3) and to a lesser degree the perineal nerve (S4), both branches of the pudendal nerve (S2, S3), provide somatic innervation of the anal canal below the dentate line. This innervation is responsible for the pain felt during thrombosis of external haemorrhoids as well as pain after surgical excision.

Above the dentate line, there is an absence of somatic innervation and only the presence of sensation provided by visceral afferent fibres which join the inferior hypogastric plexus. Hence, internal haemorrhoids are only painful if acutely prolapsed or thrombosed.

Aetiology

Theories of Pathogenesis

Varicose Vein Theory

Venous dilatations within the haemorrhoidal specimens raised the concept that haemorrhoids were the result of dilatation and weakness of veins. The absence of valves in the portal system along with the upright posture of human beings was thought to result in increasing resting venous pressure. However, this theory was disputed by the discovery of portosystemic anastomosis which allows dissipation of any increased pressure [12].

Vascular Hyperplasia Theory

Haemorrhoidal tissue was believed to be cavernous tissue much like corpus cavernosum. Arteriovenous communications suggested that erectile properties of this tissue could contribute to the continence mechanism. Hence, it was referred to as corpus cavernosum recti by Virchow and Allingham and quoted by Thomson in 1975 [3]. This theory was disputed on histological grounds by Thomson who believed that the bleeding from haemorrhoids arose from ‘capillaries in the lamina propria’ rather than the venous dilatation. Though this theory was thrown into disrepute in the long run, it did highlight the arteriovenous connections, as

opposed to solely venous communications, and explained the nature of the bright red bleeding from haemorrhoids.

Infection/Inflammation Theory

Quanu and Hartmans in the nineteenth century suggested that repeated trauma by defecation resulted in infections and a consequent weakness of the wall of the haemorrhoidal vein, as stated by McGivney in 1967 [13]. However, a distinct lack of histological evidence made this theory unlikely. Further anatomical dissections revealed that these so-called venous dilatations were physiological.

Morgagni in the seventeenth century proposed that the effects of the erect posture could be maintained, until affected by infection, in which case haemorrhoids result.

With further anatomical and histological developments into haemorrhoidal aetiology, these theories were dismissed by Jackson and Robertson in the American proctologic society meeting in 1964. These modern ideas were supported by the fact that haemorrhoids histologically demonstrate venous thickening as opposed to thinning [14].

Sliding Anal Lining Theory

Detailed observation of microscopic sections of haemorrhoids from over 4,000 patients in Tennessee by Gass and Adam demonstrated loose and fragmented submucosal collagenous and connective tissue stroma as the fundamental early lesion in haemorrhoidal disease [15]. The vein walls showed a compensatory thickening rather than thinning as proposed in the varicose vein theory. This paved the way for further anatomical studies demonstrating age-related disruption and thickening of the collagen network which provides support for the anal cushions. This results in eventual sliding of anal cushions and explains the progressive nature of haemorrhoids with age.

Predisposing Factors

Analysis of predisposing factors adds to our knowledge of why haemorrhoids occur. Though many have been identified as risk factors, evidence for these remains limited (Table 13.1).

Table 13.1 Symptoms unlikely to be attributed to haemorrhoids alone

Presentation	Alternate diagnosis
Change in bowel habit	Inflammatory bowel disease, malignancy
Faecal discharge/seepage	Rectal prolapse, neoplasm, polyp
Iron deficiency anaemia	Colorectal cancer (right>left)

Diet and Stool Consistency

Many studies have analysed the connection between constipation and haemorrhoids. Early attempts on disease management concentrated on increasing fibre intake and normalising bowel habit. Parks in 1956 suggested that the hard faecal matter resulted in venous obstruction and consequent engorgement of the haemorrhoids [16].

However, a large epidemiological study by Johnson et al. in 1990 demonstrated that constipation is not associated with haemorrhoidal disease, and indeed later studies suggest that conversely there might be an association between diarrhoea and haemorrhoids [17].

Further work by Gibbons et al. has cast doubt on the theory that constipation is a contributing factor to the development of haemorrhoids. Their group of constipated women had normal anal pressure and compliance profiles [18].

Hancock did not demonstrate a difference in bowel frequency in patients with haemorrhoids but did demonstrate that patients with prolapsing haemorrhoids were more likely to strain that controls. Whether this was cause or effect of haemorrhoids remains unclear [19].

Defecatory Position

It has been postulated that protracted time sitting or squatting on the lavatory with an unsupported perineum is associated with the development of haemorrhoids [3]. However, scientific evidence for this theory remains limited.

Genetics

Though a positive family history is common in patients with haemorrhoids [20], no specific genetic breakthrough has been made as yet. Family history may be secondary to common cultural and diet-related factors as opposed to

genetics alone. In addition there may be bias in parents of the same sex who are more likely to report perianal disease than parents of the opposite sex.

Abdominal and Pelvic Causes

Physiological causes of increased intra-abdominal pressure include pregnancy. Pathological causes include uterine fibroids, abdominal tumours and ovarian cysts. Haemorrhoids may be associated with prostatism and hernias, both of which are associated with abdominal straining, although the association with hernias may be collagen related. Pressure from pelvic masses may cause engorgement of haemorrhoids by obstructing venous return.

Functional Abnormalities of Haemorrhoids

Despite advances in anatomy, and many studies on aetiology of haemorrhoids, the question remains: if anal cushions are part of normal human anatomy, what makes them transform into haemorrhoids? Advances in physiological studies may add further answers. We analyse the evidence suggesting increased anal pressure in patients with haemorrhoids below.

Increased Anal Pressure

Anorectal physiology studies demonstrate increased resting anal pressure in patients with haemorrhoidal disease. Whether this contributes to or is the result of haemorrhoids remains unclear. Whilst an increased sphincter tone may result in hypertrophy of anal cushions, constant distension of anal cushions may itself result in an increased resting anal pressure. Arabi et al. demonstrated significantly higher anal pressures in patients with haemorrhoids, but this was reserved for young male patients [21]. Anal pressures returned to normal in patients 3 months after surgical haemorrhoidectomy, yet normal resting pressures are not affected by haemorrhoidectomy [22]. Only partial reduction in pressure is noted in patients with rubber band ligation [23]. As pressures return to normal after haemorrhoid surgery, it could be argued that the increased resting anal pressures are more likely to be the result of haemorrhoidal disease rather than the cause of it.

Not all patients' haemorrhoids exhibit raised anal resting pressures. This implies that haemorrhoidal disease is multifactorial in aetiology [24]. Patients with anal fissures have high anal pressure profiles but do not necessarily develop prolapsing haemorrhoids [21]. In addition, the prevalence of haemorrhoids increases with age, whereas the anal sphincter pressure declines with age.

Studies have investigated the predilection of female gender and elevated anal pressures causing haemorrhoids. Though some reports encourage this notion [23, 24], most studies do not reach statistical significance to allow us to establish a clear connection.

External Anal Sphincter

The external sphincter of patients with haemorrhoids demonstrates muscle hypertrophy, possibly as a result of increased contraction. This has been demonstrated on EMG recording and may contribute to the raised anal canal pressure. Using single-fibre EMG, Bruck et al. demonstrated increased external sphincter fibre density in haemorrhoids patients, implying reinnervation after denervation [25]. Pudendal nerve terminal motor latency was also prolonged implying impaired conduction.

Internal Anal Sphincter (IAS)

Gibbons et al. did demonstrate an upward shift of the resting anal compliance curve in patients with prolapsing haemorrhoids. In addition maximal squeeze pressure (under conditions of maximal external sphincter contraction) and minimum residual pressures (during internal sphincter inhibition) were similar in patients with haemorrhoids and in controls [26].

Yet, Farouk et al. found no difference in the electrical oscillation frequency of the internal sphincter, and Kerremans only demonstrated abnormal spiking activity in one patient with haemorrhoids [27, 28].

Hence, even though the IAS has been linked to the increased anal pressure profile in patients with haemorrhoids, evidence is still conflicting.

Hypertensive Anal Cushions

Sun et al. investigated 20 patients with haemorrhoids and age-matched controls [29]. They demonstrated that the IAS thickness on ultrasound was not different in haemorrhoid and control patients. In addition anal pressure profiles remained high despite IAS relaxation. Pressure of the anal cushions was measured directly, and the authors postulated that increased anal cushion pressure may contribute directly to increased anal pressure.

Slow and Ultraslow Waves

Slow waves have a frequency of 10–20 per minute at rest. They tend to be more frequent in the distal anal canal. In patients with haemorrhoids, a disturbed motility gradient has been demonstrated [30]. The evidence for this is not consistent.

Ultraslow waves are fluctuations of resting anal pressure greater than 25 cmH_2O or as 10 % of baseline pressure occurring at less than two cycles per minute. They are usually eliminated by anal dilation and lateral anal sphincterotomy and persist during general anaesthesia with skeletal muscle relaxation [31]. Hancock et al. noted an increase in maximal anal pressure and the presence of ultraslow waves in patients with haemorrhoids. These ultraslow waves are also abolished 3 months post-haemorrhoidectomy [19].

Anal and Rectal Sensation and Inhibition

Patients with haemorrhoids demonstrate reduced anal electrosensitivity and temperature sensation more in the proximal and mid anal canal perhaps because of prolapse of less sensitive rectal mucosa [24].

Evidence regarding inhibition is also conflicting. Hence, no concrete statements can be made about the effect of haemorrhoids on rectoanal inhibition.

Author's Opinion

In summary, there are as yet no concrete theories on aetiology of haemorrhoids and their pathogenesis. It is likely that an inherent or acquired disorder of the connective tissue of the anal cushions, such as reduced collagen to protein ratio or abnormal collagen subtypes, and resultant thickening and disruption of the collagen/protein network, is the underlying histological and anatomical basis for haemorrhoids. The longitudinal anal sphincter is an integral component of this network acting as a dynamic musculoelastic skeleton supporting the anal cushions. Changes in protein and collagen make-up affect the contractility and normal functioning of the longitudinal sphincter fibres. This together with dietary factors, and abnormal defecatory behaviour which encourages straining, is likely over a period of time to lead to decent of anal cushions and development of haemorrhoids. Haemorrhoids then become complicated and symptomatic as a result of repeated trauma and shearing forces during defecation. The reported anomalies in anorectal physiology including internal anal sphincter pressures and pudendal nerve terminal motor latency (PNTML) are likely to be secondary to these changes rather than primary causal factors.

Presentation of Haemorrhoids

A precise history and examination are essential in making a correct diagnosis of haemorrhoids and in ensuring that concomitant anorectal pathologies are not missed. The clinician must enquire about the nature, duration and severity of symptoms and the effect that these have on the patients' quality of life. In addition, establishing a basic understanding of what the patients' expectations are is important (Table 13.2).

History

Internal and external haemorrhoids may present with different symptoms.

Table 13.2 Presentation of external and internal haemorrhoids

	Common presentation	Additional features
External haemorrhoids	Bleeding Discharge Pruritus Skin tags	Pain (only when thrombosed)
Internal haemorrhoids	Bleeding Prolapse	Pruritus

It is important when taking a history to enquire about:

1. *Age*: Patients usually present between the ages of 30–50. In younger patients, the diagnosis of inflammatory bowel disease and polyposis must be kept in mind, whilst in older patients, the possibility of carcinoma should always be considered.
2. *Pregnancy and parity*: Pregnancy and childbirth are relevant risk factors for haemorrhoids in females. Haemorrhoids during pregnancy improve after the pregnancy is over.
3. *Medical history*: Patients with bleeding disorders or on represent a higher risk of bleeding from procedures, whether in the setting of the outpatient clinic or in the operation theatre. A history of COPD or chronic cough may indicate increased abdominal pressure. Similarly patients with liver failure are more likely to have rectal varices as a result of portal hypertension.
4. *Family history*: Family history is relevant, especially to exclude familial polyposis syndromes.
5. *Bowel habit*: A thorough history of bowel habit, of both constipation and diarrhoea, and of straining is necessary. Apart from being possible risk factors, a change of bowel habit may also be a sinister presentation of bowel cancer. In the constipated patient, a history of dietary intake is relevant, as many patients need laxatives after their haemorrhoidal treatment.
6. *Lifestyle*: It is important to ascertain prolonged periods of sitting on the lavatory reading books or squatting. The latter may be associated with haemorrhoids or may cause worsening haemorrhoidal symptoms especially prolapse. Occupations involving straining as a result of heavy lifting or pushing can also contribute to the development of haemorrhoids or their deterioration. A history of anal intercourse should be taken in order to exclude other anorectal pathologies such as condylomata, fissures and anal ulcers, perianal abscesses or fistulas.
7. *Previous medical and operative treatment*: Previous operative management should be enquired about. Anal surgery in patients who have already had an internal sphincterotomy may have deleterious effect on their continence. Recurrence after haemorrhoidectomy is unusual, and repeat procedures should be performed with care in order to avoid anal stenosis.

Internal Haemorrhoid Symptoms

Bleeding

Painless, bright red bleeding on defecation is the most common presentation of haemorrhoids. Painful bleeding should suggest the diagnosis of an anal fissure. Bleeding should not be mixed into stools and is instead described by patients as dripping into the toilet bowl. The American Society for Gastrointestinal Endoscopy and the Society for the Surgery of the Alimentary Tract advise a minimum of proctoscopy and flexible sigmoidoscopy for bright red bleeding.

Further colonoscopy is indicated when the bleeding is atypical, there are no haemorrhoids on examination or there are other risk factors for colonic cancer.

Prolapse

Prolapse occurs in larger haemorrhoids and is usually painless. The degree of prolapse is taken in account in the classification of internal haemorrhoids.

First-degree haemorrhoids bleed but do not prolapse, second-degree haemorrhoids prolapse but reduce spontaneously, third-degree haemorrhoids prolapse and have to be reduced manually, whilst fourth degree haemorrhoids cannot be reduced and are classified as external haemorrhoids.

This classification system is still commonly used by colorectal surgeons, but does not take into account other symptoms and the effect of symptoms on patient's life. In particular, haemorrhoids may not bleed, but cause other symptoms, making their classification difficult. It is often therefore clinically more relevant to classify haemorrhoids more simply as internal or external haemorrhoids. By and large external haemorrhoids will require some form of surgical excisional procedure to adequately treat, whilst internal haemorrhoids can usually be dealt with by office procedures or haemorrhoidal ligation techniques.

In the case of prolapsed haemorrhoids, patients may experience a sensation of fullness or pressure, the need to defecate or incomplete emptying of their bowels. In the case of externally prolapsed haemorrhoids, seepage or soling may occur along with a palpable swelling.

Discharge, Pruritus and Perineal Hygiene

Mucus is produced by columnar epithelium above the level of the dentate line. Prolapsed haemorrhoids have mucosa that is exposed, which becomes traumatised and irritated and secretes mucus. However, mucus can also be the presenting symptom of a rectal villous adenoma, cancer, mucosal prolapse, proctitis or ectropion from previous surgery. Pruritus is often the result of mucus discharge and perineal soiling. Fully prolapsed haemorrhoids in particular can produce a lot of discharge, as well as soiling and bleeding. This can be particularly troublesome in maintaining standards of perineal hygiene resulting in perianal excoriation, fissuring and dermatitis.

Pain

Pain is an unusual feature of haemorrhoids as internal haemorrhoids lack somatic innervation. It should alert the clinician to the possibility of an alternate diagnosis.

Nevertheless, pain may be associated with haemorrhoids when traumatised, thrombosed or necrotic.

External Haemorrhoid Symptoms

External haemorrhoids are often associated with more pronounced symptoms. They can present with significant bleeding, discharge, pruritus and difficulties in maintaining perineal hygiene. Patients may be concerned by the appearance of prolapsed uncomfortable redundant tissue around the anus. Conversely, many patients with significant prolapsed external haemorrhoids have minimal or no symptoms.

Unlike internal haemorrhoids, external haemorrhoids have somatic innervation. Yet they are only associated with pain, when acutely thrombosed, ulcerated or strangulated. Patients often complain of a painful lump around their anus, which on inspection has a bluish tinge and is usually associated with sphincter spasm or hypertonia sometimes too painful to examine fully. Once the thrombus pierces through the skin, patients experience sudden relief of pain and may be left with a skin tag. Infection is rare but can be very serious including severe pelvic or portal sepsis. Tags may prove to be problematic in preserving good perineal hygiene or aesthetically displeasing for some.

Examination

General and Abdominal

Examination of the patient commences from the moment that they walk into the clinic. It is important to ascertain any signs of liver disease as rectal varices may be differential diagnosis. In addition cachexia and general wellbeing are important as patients with haemorrhoids are traditionally young and fit and these signs should warn the clinician of alternative diagnoses.

Abdominal examination should be carried out to look for any distension, abdominal or pelvic masses and dilated abdominal veins suggesting portal hypertension.

Anorectum

Examination of the anorectum should be carried out with a chaperone. Patients should be reassured that the examination should not be painful.

Position of the patient should aim to maximise exposure but retain the patient's confidence and dignity.

Position: Examination may be carried out in the prone jackknife position. Although this provides optimum exposure, it may be embarrassing for the patient and often requires a special couch. Alternative positions include the lithotomy position, which is more common in the USA, and the left lateral or Sims position. The latter has the benefit of making patients feel more comfortable, particularly relevant in the young who have not previously had an anorectal examination.

Inspection: Examination of the anorectum should commence with the inspection of the perianal skin to identify external haemorrhoids, skin tags, prolapsing internal haemorrhoids, rectal prolapse, external openings or fistulas, fissures, anal warts, dermatitis and skin pigmentation.

Palpation: Palpation should be performed to assess any external masses or induration associated with a fistula or abscess. A digital rectal examination should be carried out to assess sphincteric tone and the presence of an anal or rectal mass or blood. Pain should alert the clinician to consider an alternative diagnosis such as sepsis, ulceration or a tumour, which may require examination under anaesthesia. The patient may be asked to strain to demonstrate any rectal or haemorrhoidal prolapse.

Proctoscopy (*Anoscopy*): This should be routinely carried out in all patients with haemorrhoids. There are numerous designs of proctoscope which allow performing treatments such as rubber band ligation by virtue of a longitudinal groove.

Sigmoidoscopy: Patients should also undergo a rigid sigmoidoscopy which usually allows examination 15–20 cm into the rectum. Some units perform routine flexible sigmoidoscopy in the outpatient setting. This is particularly relevant in patients where a left-sided colorectal cancer is suspected from the history. Red flag symptoms should always prompt a formal investigation by colonoscopy.

Examination on the commode or toilet: When prolapse is the predominant symptom, patients must be examined whilst straining in the sitting position as only this examination will accurately distinguish between haemorrhoidal prolapse versus mucosal rectal prolapse and full thickness rectal prolapse. Unfortunately all too often this examination is omitted, and patients undergo inappropriate or inadequate treatment for their presenting symptoms due to inaccurate diagnosis of the extent of the prolapse.

In general, young patients (under 40 years of age) with symptoms of prolapse, pruritus and discharge compatible with haemorrhoids should only require proctoscopy and rigid sigmoidoscopy. Rectal bleeding should be investigated with a minimum of flexible sigmoidoscopy in the young patient with no red flag symptoms. In older patients and those with red flag symptoms, or symptoms that are not explained by clinical examination, a formal investigation of the rest of the colon should be carried out.

References

1. Mason P. Napoleon's haemorrhoids: and other small events that changed history. Philadelphia: Skyhorse Publishing; 2009.
2. Milligan ETC, Morgan CN, Jones LE, et al. Surgical anatomy of the anal canal and the operative treatment of haemorrhoids. Lancet. 1937;2:1120–4.
3. Thomson WHF. The anatomy and nature of piles. In: Kaufman HD, editor. The haemorrhoid syndrome. Tunbridge Wells: Abacus; 1981. p. 15–33.
4. Johnson FP. The development of the rectum in human embryo. Am J Anat. 1914;16:1–57.
5. Lestar B, Pennickx F, Rigauts H, et al. The internal sphincter cannot close the anal canal completely. Int J Colorectal Dis. 1992;7(3):159–61.
6. Lestar B, Pennicinckx F, Kerremans R. The composition of anal basal pressure. An in vivo and in vitro study in man. Int J Colorectal Dis. 1989;4:118–22.
7. Read MG, Read NW, Haynes WG, et al. A prospective study of the effect of haemorrhoidectomy on sphincter function and faecal continence. Br J Surg. 1982;69:396–8.
8. Haas OA, Foc Jr TA. Age related changes and scar formations of perianal connective tissue. Dis Colon Rectum. 1980;23(3):160–9.
9. Willis S, Junge K, Ebrahimim R, et al. Haemorrhoids – a collagen disease? Colorectal Dis. 2010;12(12):1249–53.
10. Murray-Lyon IM, Kirkham JS. Hepatic abscesses complicating injection sclerotherapy of haemorrhoids. Eur J Gastroenterol Hepatol. 2001;13(8):971–2.
11. Gray H. Chapter 63: Vascular supply and lymphatic drainage of the perineum. In: Anatomy of the human body. 20th ed. Philadelphia: Lea and Febringer; 1918.

12. Bernstein WC. What are haemorrhoids and what is their relationship to the portal venous system? Dis Colon Rectum. 1983;26:829–34.
13. McGivney J. A re-evaluation of etiologic factors of haemorrhoidal disease. Ariz Med. 1967;24:333–6.
14. Jackson CC, Robertson E. Etiologic aspects of haemorrhoidal disease. Dis Colon Rectum. 1965;8:185–9.
15. Gass OC, Adams J. Hemorrhoids: etiology and pathology. Am J Surg. 1950;79(1):40–3.
16. Parks AG. Surgical treatment of haemorrhoids. Br J Surg. 1956;43(180):337–51.
17. Johanson JF, Sonnenberg A. The prevalence of hemorrhoids and chronic constipation. An epidemiologic study. Gastroenterology. 1990;98:380–6.
18. Gibbons CP, Bannister JJ, Read NW. Role of constipation and anal hypertonia in the pathogenesis of haemorrhoids. Br J Surg. 1988;75:656–60.
19. Hancock BD, Smith K. The internal sphincter and Lords procedure for haemorrhoids. Br J Surg. 1975; 62:833–6.
20. Loder PB, Kamm MA, Nicholls RJ, Phillips RK. Haemorrhoids : pathology, pathophysiology, and aetiology. Br J Surg. 1994;81:946–54.
21. Arabi Y, Alexander-Williams J, Keighley MRB. Anal pressures in haemorrhoids and anal fissure. Am J Surg. 1977;134:608–10.
22. Creve U, Hubens A. The effect of Lord's procedure on anal pressure. Dis Colon Rectum. 1979;l22:483–5.
23. Deutsch AA, Moschkovitz MM, Nudelman I. Anal pressure measurements in the study of haemorrhoid etiology and their relation to treatment. Dis Colon Rectum. 1987;30(11):855–7.
24. Read NW, Bartolo DCC, Read MG, Hall J, Haynes WG, Johnson AG. Differences in anorectal manometry in patients with haemorrhoids and patients with the descending perineum syndrome: implications for management. Br J Surg. 1983;70:656–9.
25. Bruck CE, Lubowski DZ, King DW. Do patients with hemorrhoids have pelvic floor denervation? Int J Colorectal Dis. 1988;3:210–4.
26. Gibbons CP, Trowbridge EA, Bannister JJ, et al. Role of anal cushions in maintaining continence. Lancet. 1986;1:886–8.
27. Kerremans R. Electrical activity and motility of the internal anal sphincter. Acta Gastroenterol Belg. 1968;31:465–82.
28. Farouk R, Duthie GS, Mac Gregor AB, et al. Sustained internal sphincter hypertonia in patients with chronic anal fissure. Dis Colon Rectum. 1994;37(5):424–9.
29. Sun WM, Peck RJ, Shorthouse AJ, et al. Haemorrhoids are associated not with hypertrophy of the internal anal sphincter, but with hypertension of the anal cushions. Br J Surg. 1992;79:592–4.
30. Kerremans R. Morphological and physiological aspects of anal continence and defecation. Brussels: Arscia; 1969.
31. Schouten WR, van Vroonhoven TJ. Lateral internal sphincterotomy in the treatment of haemorrhoids. A clinical and manometric study. Dis Colon Rectum. 1986;29:869–72.

Operative Management

14

Antonio Longo, Adolfo Renzi, and Francesco Prete

Introduction

To be effective, surgical treatment must be based on detailed knowledge of the nature of the pathology that it aims to resolve. Therefore, the deeper and more accurate the knowledge of anatomy, physiology and pathogenesis, the higher the chances of being able to develop surgical techniques that are rational and focused on a specific disease.

Haemorrhoidal disease has been known for thousands of years for its high incidence among the human species and the relative ease with which it is diagnosed. Even the Bible mentions how God punished the Philistines making them the target of an epidemic of haemorrhoids, although this probably has to be attributed to an incorrect translation.

In the course of the millennia, countless theories have followed one another trying to clarify the anatomical and physiological nature of haemorrhoids and the dynamics of their causes and pathogenesis. Without indulging in details, which would go beyond the scope of this chapter, it seems appropriate to briefly report some concepts of anatomy and physiopathology to better understand the rationale of the various therapeutic options for the haemorrhoidal disease.

Today it is widely recognised that the haemorrhoidal cushions play a role in anal continence because of their ability to inflate and deflate rapidly [1]. This ability to adjust their volume is due to the anatomical nature of the cushions, with their numerous arterial and venous shunts that produce vascular lacunar spaces. The blood supply to the haemorrhoidal cushions through the superior, middle and inferior haemorrhoidal arteries, which undoubtedly exceeds the sole biological needs, has the ultimate purpose of allowing the haemorrhoids to quickly fill with blood to optimise, in synergy with the anal sphincters, the anal continence. It is thus a case of "hyper-vascularisation" that supports the functional role optimising the anal continence.

The haemorrhoidal cushions are kept in their position by connective tissue and smooth muscle fibres [2] and are covered with anal mucosa. The anal mucosa overlying the haemorrhoids, besides being arranged in longitudinal folds that provide for an adequate aperture of the anal canal during defecation, is specialised in the discrimination of rectal contents and therefore is an anatomical structure with a fundamental role regarding the anorectal reflexes and, thus, regarding anal continence. These simple considerations have led to two important reflections.

The first is that the scope of "hyper-vascularisation" is that of guaranteeing a hyperflow of blood to the haemorrhoidal vessels. This aspect, which some, as we will later see,

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have seen as one of the causing factors of the haemorrhoidal disease, is instead a physiological peculiarity that exists with the only aim to quickly increase haemorrhoidal volume to seal the anus.

The second reflection is that ablation or destruction of the haemorrhoids, regardless of the methods, ends up in weakening – in various degrees – anal continence and dilatability.

Among the many theories on the pathogenesis of the haemorrhoidal disease that have followed one another in centuries, some have obviously left such a long-lasting impression that they are still evoked nowadays, often wrongly, in therapeutic decisions. Among these theories we find the so-called theory of the varicose veins, dating from age of Galen and Hippocrates [1]. Going back to the observation of the frequent association between dilatation of the haemorrhoidal plexuses and symptoms, this theory, although identifying different causative factors for haemorrhoidal dilatation, ended up equating haemorrhoidal disease to rectal varices, like the ones following portal hypertension. Subsequently it has well been clarified that rectal varices caused by portal hypertension are a rare pathology and distinct from that of the haemorrhoids. As the dilatation of the haemorrhoidal veins is always associated to a prolapse, it is fundamental to understand the pathogenetic correlation between these two phenomena. Angiographic studies have demonstrated that haemorrhoidal prolapse causes a venous kinking between the middle and inferior haemorrhoidal veins and a stretching of the superior haemorrhoidal vessels that obviously obstacle outflow. This obstacle is worsened by sphincter hypertone. These factors can create a venous dilatation with blood stagnation, thrombosis and oedema. It is therefore evident that venous dilatation is a complication following prolapse and not a primary cause of haemorrhoidal pathology.

Although nowadays this theory is no longer supported among proctologists, the idea that the haemorrhoidal pathology is similar in some way to varicose veins continues to be widespread. This is proved by the fact that many drugs prescribed for haemorrhoidal pathology are the same used for lower limb varices. The good business for the pharmaceutical companies producing the so-called vessel protectors and vasoactive drugs and the natural inclination of patients to try whichever form of treatment just to avoid surgical treatment, which has always been considered an extremely painful experience, seem to be among the reasons for the survival of this theory.

The theory of the so-called vascular hyperplasia, particularly popular in Europe, probably has its origin in a certain histological similarity between the prolapsed haemorrhoidal cushions and angiomatous tissues. Although it has been abandoned, this theory deserves to be remembered here as many of the studies performed to prove its validity have contributed to clarify the anatomical structure and the physiological function of the haemorrhoids. In any case it has been clearly demonstrated histologically that haemorrhoidal specimens show no signs of tissue hyperplasia.

The theory nowadays largely accepted by proctologists is the one proposed by Gass and Adams in 1950 [3], *the sliding anal lining theory*. This theory assumes the prolapse of the anal lining as the pathogenetic cause of haemorrhoidal disease. It is based on the concept that "fragmentation" of the ligaments of Treitz and Parks, which support the haemorrhoidal cushions, causes prolapse of haemorrhoids and of anal mucosa. The prolapse is considered a predisposing cause of all the haemorrhoidal symptoms and complications.

It is useful making some considerations on Goligher's classification [4] that, as is known, is the most widespread tool to assess haemorrhoidal pathology. Although this classification, that divides haemorrhoidal prolapse into four grades, is accepted unanimously, some of its elements have to be viewed critically. The definition of the first degree of prolapse, for example, is not entirely clear, as it includes haemorrhoids that are "not prolapsing, but increased in volume, and projecting into the anal canal and bleeding". It is evident that this definition of first degree is bound to be considered critically for several reasons: (a) it is not very clear why you would include non-prolapsed haemorrhoids in a classification based on prolapse itself; (b) besides the fact that projection into the lumen of the anal canal is a normal anatomical condition, the concept of

"increased volume" appears unclear as no reference is made to either what a normal volume is or to a range of normality; it is well known that haemorrhoidal volume is very variable in the population at large and even in the same subject depending on physiological conditions; and (c) in order to define this first-degree prolapse, a symptom like bleeding is being used which is illogical in a classification otherwise based on the clinical behaviour of the prolapse (whether reducible spontaneously, manually or non-reducible). This incongruence in classification is probably due to the fact that the way the theory of prolapse was formulated is not able to sufficiently explain the clinical events and, in a certain way, the true essence of haemorrhoidal pathology.

Moreover, there is much confusion regarding the significance of the prolapse degree even among specialists – although Goligher cannot be blamed for this. It is not unusual to read endoscopic reports defining the grade of prolapse based on an evaluation of the volume of the inspected haemorrhoidal cushions rather than an evaluation based on history and clinical appearance: in fact a distinction between second and third degree can only be made by questioning the patient (whether the prolapse retracts spontaneously or requires manual manoeuvres). Grade IV is the only one that can be diagnosed by a doctor during proctological exam.

The three pathological theories outlined, even if conceptually different, have some elements in common. Varices, vascular hyperplasia and prolapse generated by wearing of supportive tissue are considered irreversible anatomical and histological alterations. Moreover, all the variations in pathogenesis delineated so far tend to indicate haemorrhoidal pathology as a primary disease, with cause and effect limited to the anal canal. Because of these convictions, the elimination of haemorrhoidal tissue was considered, until recent times, the only logical and effective therapeutic treatment. We will see that it cannot be that way.

For reasons of better readability of this chapter, we will progressively analyse the solutions that were proposed in the course of time for treatment of haemorrhoidal pathology, reporting techniques and results. We will first take into consideration outpatient procedures that, as we will see, are mainly focused on destroying haemorrhoids through activation of tissue necrosis, even though in different ways. At the end of this section, dearterialisation will be discussed. After that we will analyse the actual surgical techniques for removal of haemorrhoids and finally, following the presentation of the unitary theory of rectoanal prolapses, the techniques stapled haemorrhoidopexy (SH) and stapled transanal rectal resection (STARR).

Ambulatory Treatment

Sclerotherapy

This outpatient procedure consists in injecting chemical agents in the haemorrhoidal plexuses with subsequent fibrosis, scarring, shrinking and fixation of the haemorrhoids. These effects are secondary to the obliteration of haemorrhoidal vessels induced by the sclerosing agent solution.

The first attempt at obliterating haemorrhoids through the use of sclerosing injections was performed by Morgan, who in 1869 used a solution of ferrous persulphate to treat external haemorrhoids. In 1871 sclerotherapy with phenol and other chemical agents was introduced in the United States and a few years later indicated as "cure for haemorrhoids without pain or surgery" [5].

The absence or at least the scarce availability of anal specula at that time made only prolapsed haemorrhoids treatable that were attacked with a massive injection of sclerosing solution. The enthusiasm that this technique inspired can be imagined when thinking that even Mr. Andrews [1], then president of the Chicago Medical Society, considered this technique appropriate despite an investigation he had conducted himself on over 3,000 patients undergoing sclerotherapy (many of them treated by travelling charlatans and inexperienced doctors) and which revealed a high number of complications, including severe pain and even nine deaths. Andrews indicated both the use of phenol as sclerosing agent and bed rest for at least 8 h following

treatment as indispensable elements for a good outcome of the therapy.

In this same period also, Kelsey in the United States and Edwards in England recognised the therapeutic efficacy of sclerosing treatment. These authors found that the incidence of complications were lower when a less concentrated solution of phenol, between 5 and 7.5 %, was used [6, 7].

We have to give credit to Terrell for being the first to identify the most appropriate indication for sclerosing therapy in the treatment of internal haemorrhoids. In one of his publications in 1913, he reported on the brilliant results he obtained and with this contributed significantly to the spreading of this method [8].

In the following decade, Anderson published an interesting historical review, in which he reports on what has happened since then, with particular attention to the historical and not only the scientific implications [9].

Concerning sclerosing agents, it should be remembered that in the last 140 years, i.e. from the first injections performed by Morgan, various combinations have been proposed, very often based on alchemistic rather than truly scientific approaches. The 5 % phenol solution in almond or vegetable oil proposed since the beginning of the twentieth century though has asserted itself and to date is still the most used sclerosing agent in the world and in fact the only one used in the United Kingdom.

Indications

Internal non-prolapsing haemorrhoids are the ones that benefit most from sclerotherapy. Although occasionally a single, slightly protruding haemorrhoid plexus can be treated successfully, more commonly, in case of voluminous haemorrhoids accompanied by abundant submucosal tissue and requiring manual repositioning after defecation, sclerotherapy is completely ineffective. Also, such a procedure is not to be performed in cases of internal haemorrhoids associated with thrombi and anodermal ulcers. External haemorrhoids constitute an absolute contraindication, as treatment would not only be ineffective but also cause even more severe pain and sloughing.

Technique

Sclerotherapy is usually performed with the patient in jackknife or Sims position. Following insertion of an anoscope and identification of the haemorrhoidal plexuses that need treatment, the surgeon goes on to inject the sclerosing solution. Use of local antiseptic solutions or analgesics is not necessary. It is particularly useful to have a needle with an angled distal end that facilitates vision through the anoscope. In the absence of a dedicated needle, a spinal anaesthesia needle can be used efficiently too. The needle tip is inserted into the centre of the haemorrhoidal plexus, while avoiding going down too much towards the pectinate line. A single injection can be made in each plexus to be treated. The formation of a submucosal bulge indicates a correct execution of the manoeuvre. Depending on the type of solution, different quantities of solution can be used: with regard to the ones available on the market, based on sodium morrhuate, quinine, urea hydrochloride or Sotradecol, generally no more than 3 ml is used in total; in the case of solutions with 5 % phenol in oil, it is possible to use 3 ml for each haemorrhoidal plexus treated.

Results

In the last few years, only a few studies have been published, at least in English. In 1985, Khoury reported the results of a randomised study aiming at comparing efficacy and safety of single vs. multiple injections, showing that a single session of sclerotherapy (with more than one plexus treated) is as effective as a treatment consisting of multiple sessions [10].

In a study by Dencker [11], sclerotherapy resulted scarcely effective if compared to internal haemorrhoid ligation or to the excision as described by Milligan. Meagre long-term results, at 4 years, are also reported by Santos, who describes the technique as effective only in the short term [12]. Also Alexander-Williams came to the same conclusions in 1975 [13].

Unfavourable outcomes of sclerotherapy also emerge from other studies, e.g. those published by Ambrose et al. and Gartell et al. [13, 14], in which sclerotherapy is compared, respectively, to photocoagulation and to rubber band ligation.

Concerning complications, the most frequent one is sloughing which however can almost always be ascribed to an incorrect technical performance of the procedure with a too superficial or too abundant injection or to a second injection done too soon after the previous one. Other, less frequent complications, which are however typical of the method used, are necrosis and haemorrhoidal thrombosis, the treatment of which is always conservative. Cases of "chemical stenosis" of the anal canal [15] have also been described for which anoplasty may be necessary.

Development of a burning sensation in the anal canal is a late complication which, although uncommon, is seen following repeated sclerotherapies. This complication is only partially responsive to topical medications, and its symptoms can be very debilitating.

Also episodes of bacteraemia may represent a complication of sclerotherapy. In 1981 Adami et al. [16] observed this complication with a frequency of 8 %; although this almost never leads to a sepsis, prophylactic antibiotic therapy is indicated, especially in subjects at risk.

Other very rare complications, such as urological sepsis, prostate and seminal vesicles abscess, epididymitis, uro-perineal fistula, necrotising fasciitis involving the perineum and scrotum, rectal perforation and septic shock, were described by both Ribbans and Radcliffe [17] in 1985 and Guy and Seow-Cohen [18] in 2003.

Rubber Band Ligation

This is an outpatient procedure for the treatment of haemorrhoidal pathology based on the effect of fixation-necrosis determined by ligation of the haemorrhoidal plexuses with elastic rings. Although the original idea goes back to Blaisdell [19], the description of the general technical principles and the development of a dedicated instrument are the work of Barron who, in 1963, in two scientific papers published within a few weeks, reported the satisfactory results obtained in 150 patients, almost all of them treated as outpatients [20, 21].

Currently rubber band ligation is one of the most widely used outpatient procedure in the United States. In the fifth edition of his textbook, Corman [22] states that the results of this procedure are so gratifying that they have induced him to prefer this technique over surgical haemorrhoidectomy in about 80 % of his patients.

Indications

This technique is usually indicated in first- and second-degree haemorrhoids.

Technique

This technique can be performed safely with the patient in different positions, without anaesthesia and with the optional use of an enema. After introducing an anoscope, the physician proceeds by placing one or more often two elastic bands (in case one breaks) at the base of the haemorrhoidal plexus to be treated, on which he previously applied gentle traction; this causes strangulation of the blood vessels and cuts off the blood supply to the which eventually falls off after 5–7 days. The remaining small ulcer heals off if the mucosa fixed on the underlying muscular layer is left in place.

The original technique by Barron implies the use of an instrument designed by himself, consisting of a simple metallic rod with a hollow cylinder fixed at its end whose main axis runs parallel to that of the metallic rod; some elastic rings are mounted on this cylinder. It is a bimanual manoeuvre: with one hand the physician leads the distal end of a grasping forceps through the cylinder that he holds in place with his other hand placed on the rod; with the forceps he grasps the haemorrhoidal plexus and drags it through the cylinder from which the elastic bands are released by means of a sliding mechanism.

The biggest haemorrhoidal plexus is treated first, taking care that the elastic bands are positioned 1–3 cm cranially to the dentate line. Although it is appropriate to treat only one nodule per session, it is not uncommon that bands are applied on more than one plexus simultaneously.

Nowadays different instruments can be used to perform problem-free elastic ligations. Also suction devices as the McGown and Lurz-Goltner

instruments are available which use the force of an aspirator instead of a forceps to pull the haemorrhoidal tissue into the cylinder. The advantage offered by this type of instruments is that it is possible to perform the ligation procedure with one hand only, while the other hand holds the anoscope: it is therefore not necessary then having an assistant. When compared to those requiring manual traction, however, these instruments have a disadvantage in that the cylinder is generally smaller, so less haemorrhoidal tissue is involved in each ligature. Among manual traction instruments, the McGivney ligator is particularly appreciated for the possibility to adjust the position of the cylinder and handle with respect to the rod thanks to a system of joints that can be rotated 360°, thus always guaranteeing an optimal vision for the surgeon.

More recently a single-use instrument, the O'Regan System, was launched. Apart from the single-use design of the device, its main advantage is in the suction modality, which is generated with a system similar to a syringe that eliminates the need for tubes and aspirators.

For some days after the procedure patients can experience tenesmus that usually is easily controlled with warm washes and/or common analgesics. Causing the patient constipation should be avoided as it has been reported to worsen results [23].

Results

The reported success rate of this method varies depending on the length of follow-up, grade of haemorrhoidal pathology and the criteria used to define success and failure [24–27]. In the majority of cases, two-thirds to three-quarters of patients with either first- or second-degree haemorrhoids respond positively to rubber band ligation, although in a significant number of these patients repeating the procedure is necessary [28].

In a study conducted by Savioz et al. [29] on 92 patients, aiming at detecting the rate of recurrence, the author reports that 23 % of patients needed a new session of ligation at 5 years and 32 % at 10 years.

From analysis of a sample of almost 3,000 patients undergoing rubber band ligation, followed for 12 years, Bayer et al. [30] states that 79 % did not need any further therapy, 18 % underwent a new session of rubber band ligation and 2.1 % were treated with surgical haemorrhoidectomy because of persistence of symptoms.

As already noted pathological haemorrhoidal plexuses can be ligated in one or more sessions. In an interesting retrospective study, Lee et al. [25] compares the results of sessions with single band ligation with those of sessions with multiple ligations. The author identifies multiple ligation sessions as a cause of increased patient discomfort (29 % vs. 4.5 %) and a higher incidence of vagal symptoms (12.3 % vs. 0 %). This opinion is not shared by other authors who do not detect a significant increase of morbidity when sessions with multiple ligatures are performed [28, 31–35].

Possible complications of rubber band ligation of haemorrhoids are pain, thrombosis and pelvic and perineal sepsis. The most frequent complication is pain, reported in 5–60 % of patients [34–37]. Pain is generally mild to moderate and controllable with warm local washes and analgesics. Severe pain is less frequent and often caused by positioning the elastic bands too close to the dentate line; in these cases early removal of a ligation is the only solution. Thrombosis of the internal haemorrhoids or more often of the corresponding external ones (2–3 %) is far less frequent. In these cases warm washes and painkillers can facilitate spontaneous resolution, whereas surgical excision is rarely indicated [22].

Pelvic and perineal sepsis is an infrequent but very dramatic adverse event after rubber band ligation. Documented for the first time in 1980 by O'Hara [38], it has since been reported also by other authors [39–44]. It seems that young men are the "ideal" subjects for this complication, presenting with progressive increase in anorectal and then perineal pain, with worsening difficulty in micturition, scrotal swelling and fever. Aetiology remains uncertain. Treatment obviously requires admission, often intensive care, massive antibiotic therapy, surgical debridement and possibly hyperbaric oxygen therapy.

In 1993 Bat et al. [45] published a prospective study conducted on 513 patients with the aim of detecting complications from rubber band ligation. In this series 4.6 % of patients developed minor complications including pain, dislocation of the rubber band, mucosal ulceration, priapism and urinary retention. Serious complications requiring admission, such as massive haemorrhage, severe pain and perianal sepsis, occurred in 2.5 % of patients.

Cryotherapy

Cryotherapy is based on the concept that in a tissue undergoing rapid freezing, intracellular water crystallises, cell membranes are destroyed and cell necrosis sets in. Freezing also destroys nerve terminations and induces immediate anaesthesia so cellular destruction is painless.

In the early 1970s, this procedure spread fairly rapidly and, as it had happened a 100 years before with Morgan's sclerotherapy, was indicated by various authors [46–50] as "effective and painless". However, the volume increase of the surviving adjacent cells and the oedema occurring with the increasing temperature in the tissue, together with a considerable increase in cellular tension, are the cause of a profuse serous secretion and pain. Secretion is particularly abundant if treatment is performed distally to the dentate line with the aim of treating external haemorrhoids or anal tags, with some patients being obliged to change pads constantly in order to avoid surrounding skin maceration. For this reason after the first experiences with "expanded" indications, treatment with cryotherapy is reserved only to internal haemorrhoids.

Technique

The patient is placed in Sims or jackknife position. With the aid of a plastic speculum (this material does not conduct cold), the cylindrical cryoscopic probe is positioned in contact with the part to treat or the whole length of the haemorrhoidal plexus. Time of contact is about 2 min but can vary depending on the type of probe adopted. Carbon dioxide probes, the most widely used ones due to their relatively low cost, have a lower cooling power (−89 °C)) than the much more expensive ones based on liquid nitrogen (−196 °C). The advantage of the latter is exclusively in the higher speed with which they generate the freezing of a haemorrhoidal plexus, reducing procedure time. Freezing, i.e. the transformation of the haemorrhoidal plexus into an "ice ball", occurs at −22 °C and is therefore obtainable with both types of probe. Once 2–3 min has elapsed, the probe is removed, and in a few minutes the tissue defrosts returning apparently identical to its initial state. Some authors, among them Kaufman [51], suggest a second application after a defrosting period of 5–10 min.

Results

Between the late 1970s and the early 1980s, Kaufman [51], Southam [52] and Berry and D'Acosta [53] reported very convincing results in their studies, with a degree of patient satisfaction varying from 75 to 97 %.

Subsequently, both because of longer follow-up and the enrolment of patients in wider case series and in randomised studies, the results of this technique appeared gradually less brilliant: Smith et al. [54], in a randomised study of cryotherapy vs. haemorrhoidectomy, identifies the latter as a technique which offers faster healing without residual disease. Besides describing the frequent presence of bothersome signs left on skin and thrombosis in the haemorrhoidal vessels not involved in cryo-destruction, the author also reports cases of incontinence apparently due to damages to the internal sphincter because of the impossibility of limiting the depth of cryo-generated necrosis.

In addition O'Callaghan et al. [55] and Goligher [56] both refer to the procedure as time-consuming and too frequently associated with foul-smelling and profuse secretion, irritation of perianal skin and severe pain. These authors also report of how inappropriate applications can cause stenosis and/or incontinence due to sphincter involvement.

Keighley and Williams [57] are straightforward in describing cryotherapy as a painful

therapy frequently associated with profuse secretion and sick leave for at least 1 week. These authors state that only 50 % of patients are fully satisfied, and long-term results are little predictable. Generally, this procedure is no longer recommended.

Photocoagulation

Photocoagulation is a therapeutic technique based on the possibility of converting infrared light into heat, causing coagulation of tissue proteins, water evaporation, eschar formation and subsequent scarring and fixation of the mucosa above the haemorrhoids to the muscular wall. The technique involving infrared coagulation of small haemorrhagic areas was developed by Nath et al. [58] in 1977, with the aim of obtaining haemostasis without tissue adhesion typical of diathermic coagulation. Nieger [59], in 1979, was the first to adopt this technique in the elective therapy of haemorrhoidal disease.

Technique

After inserting a proctoscope, preferably oblique cut, the infrared light generated by a halogen lamp is applied on haemorrhoidal tissue through an instrument similar to a gun that focuses light rays through a quartz piece. Three to five impulses of light of the duration of 1–1.5 s are directly applied onto haemorrhoidal tissue, without the need for anaesthetic injection for applications above the dentate line. Whether he wants to treat one or more plexuses in the same session is left to the discretion of the surgeon. The burnt tissue reacts in the same way as that destroyed through freezing (cryotherapy) or strangulation (band ligation). Healing of the respective ulcer usually occurs in a month. Further treatments can be repeated after 2–3 weeks.

Results

When compared to other techniques of haemorrhoidal "destruction", infrared coagulation offers comparable efficacy but with less complications. In a recent randomised study against rubber band ligation, published in 2006 and conducted on a sample of 94 patients, Marques et al. [60] identifies a higher frequency of haemorrhage and higher posttreatment pain with rubber band ligation. Similar results had also been reported by Ambrose et al. [37] on a sample of 268 patients. Weinstein et al. [61] indicates a higher frequency of haemorrhoidal thrombosis and late haemorrhage in his comparative study of infrared photocoagulation and rubber band ligation.

Monopolar Diathermy

Electrotherapy with direct current, or monopolar diathermy, is a technique of haemorrhoidal tissue destruction obtained through the heat generated by monopolar current.

Technique

The technique involves the use of an instrument; the most widely used example of which is the Ultroid (Microinvasive, Watertown, MA.), which releases monopolar current through a probe. After positioning of a proctoscope, the tip of the probe is placed in contact with the rectal mucosa at the apex of the haemorrhoidal plexus. The current, the intensity of which is set to the maximum tolerated by the patient, is applied for about 10 min [63–66]. Generally only one application is made per session, also because of the rather long duration of each application.

Results

Norman et al. [67] concludes from the analysis of results of a study conducted on 120 patients, that, although in over 20 % of patients further applications were necessary, this technique guarantees complete success, without complications, even in patients with third- and fourth-degree haemorrhoids.

Dennison et al. [68] pointed out the excessive length of this procedure. This author reports similar results to those obtained with monopolar diathermy and other outpatient procedures but denounces the considerable duration of each single treatment, particularly unpleasant for the patients and even the physicians themselves; this

aspect probably contributed to the scarce spreading of this technique.

Bipolar Diathermy

Bipolar diathermy (BICAP; Circon ACMI; Stamford, CT) is another technique that uses heat to cause destruction of haemorrhoidal tissue with subsequent ulceration, fibrosis and fixation of mucosal tissue to the underlying layers. This methodology of application of electrical current to tissues is supposed to limit the depth of damage in contrast to what happens with monopolar current, photocoagulation and laser therapy [62, 63].

Technique

This technique, originally developed to treat upper GI ulcers, is based on the use of dedicated forceps connected to a generator and operated by a pedal command. The heat is produced by the passage of electric current through adjacent electrodes situated at the tip of the forceps. A few instants after the application, a whitish clot forms that extends in depth for about 3 mm. All the haemorrhoids can be treated in a single session and generally no anaesthesia is necessary.

Results

Different studies have been conducted that compare bipolar diathermy with other outpatient treatments. Randall et al. [63] and Hilton et al. [64] have compared bipolar diathermy with treatment by Ultroid (monopolar diathermy): both procedures turned out to be equally effective, but due to the shorter duration of the procedure involving the use of bipolar diathermy, the latter was better accepted by the patients.

Advantages as a result of less deep penetration of the heat, and hence of less collateral damage, as well as with regard to the possibility of treating several plexuses in a single session, were reported by Hinton [69] and Dennison et al. [70] in a comparison between bipolar diathermy and photocoagulation.

Looking at bipolar diathermy in comparison with rubber band ligation has been the focus of a study by Griffith et al. [71], which did not point out any particular advantage of either technique over the other.

Finally, less postoperative pain was highlighted by Yang, in a study that compared the bipolar technique and Ultroid (monopolar diathermy) [72].

Comment on Outpatient Treatment of Haemorrhoids

Destroying haemorrhoidal tissue means accepting a theory of pathogenesis which is based on the assumption that the haemorrhoidal tissue itself is affected by primary alterations causing symptoms. Almost always the only symptom taken into account is bleeding. What emerges from the vast literature on the topic is that symptoms like soiling and itching are almost never taken into consideration, as they are considered to be independent of haemorrhoidal disease. Hence, the efficacy of these techniques is almost always judged in relation to the resolution of bleeding. When this symptom is solved, a treatment is considered effective.

In the best of scenarios, destruction of haemorrhoidal tissue implies fixation of the residual tissue and of anal mucosa, certainly not correction of the prolapse. Therefore, these techniques should not be indicated in haemorrhoidal prolapse, and in fact they are often used for bleeding internal and non-prolapsed haemorrhoids, especially cryotherapy and sclerotherapy.

We know that bleeding originates from the submucosa and that in rare cases of mucosal ulceration haemorrhoidal tissue may bleed, too. So destroying the haemorrhoidal cushions is not rational. This type of therapy is believed to be indicated in swollen haemorrhoids protruding into the lumen! It has to be noted that haemorrhoids always protrude into the anal canal lumen, and their supposed increase in volume is obviously a completely arbitrary and illogical judgement lacking any reference parameter whatsoever, given the physiological ability of the haemorrhoids to adapt their size.

The only acceptable hypothesis is a bulge which is always temporary and caused by

impeded extra-haemorrhoidal venous outflow, but also in this case it would be a therapy aimed at the effect and not the cause of a symptom.

Finally, a consideration which is certainly not shared by those who consider the destruction of haemorrhoidal tissue an appropriate treatment: what conceptual and technical revolution is that supposed to be, compared to what has been around since the life and times of Hippocrates? From using a hot iron to thermal ablation, cryotherapy, etc.? Of course Hippocrates used to treat prolapsed haemorrhoids so at least the indication was correct! In terms of its biological outcome, the effect caused by the use of the red-hot iron hook is comparable to diathermy, cryotherapy, etc. So the only revolution and evolution so far has probably taken place in the fields of anaesthesiology and analgesics and not with regard to the chemical or physical means used to destroy haemorrhoidal tissue. Given the indications and sometimes serious complications of these procedures, a critical review of their use may be necessary.

Some critical observations need to be made with regard to rubber band ligation (RBL). Blaisdell [19] and Barron [20, 21] indicated RBL as an ideal outpatient procedure to destroy internal prolapsed haemorrhoids by means of necrosis. The elastic band positioned above the dentate line needed to include internal haemorrhoid. If we agree that prolapsed haemorrhoids are not longer than usual, on the contrary, they are often shorter as they are no longer stretched by longitudinal support fibres, it is inevitable that the positioning of a rubber band causes dragging of rectal mucosa towards the dentate line. If the procedure is repeated on all of the three cushions, the anal canal will eventually be lined almost completely with mucosa. In other terms, this would be equivalent to creating an ultra-low anastomosis that in turn causes wet anus, soiling and impairment of the discrimination capacity, not to mention a certain degree of stenosis. However, quite strangely such complications are almost never mentioned and the positive results reported in recent years in the literature but above all in congresses and by American speakers reach the amazing rate of 90–95 %. In truth, without overtly admitting it, many colleagues have converted to the theory of the muco-haemorrhoidal prolapse, and instead of applying the rubber band on the haemorrhoid, they position it across the mucosa lying above, thus performing – in a much less effective manner – a haemorrhoidopexy. Far from wanting to make any accusations, it turns out that the reported success rate for RBL is much higher in those countries where insurance reimbursement for this outpatient procedure is almost equivalent to that paid for a haemorrhoidectomy for which the patient is admitted to hospital.

Haemorrhoidal Artery Ligation: HAL, DG-HAL, THD, and HAL-RAR

This technique known under the acronym of HAL (haemorrhoidal artery ligation) is a non-invasive surgical procedure for the treatment of haemorrhoid pathology developed and proposed in 1995 by the Japanese surgeon Morinaga et al. [73]. As we already had a chance to point out in the introduction to this chapter, the theoretical starting point for this approach is based on the assumption that the aetiopathology of the haemorrhoidal pathology lies in an excessive influx of blood into these structures. The procedure therefore implies the precise identification, under Doppler guidance, of the terminal branches of the superior haemorrhoidal arteries and their ligation. Various centres in Europe and in America have adopted this technique with some minimal variations and using different names: Doppler-guided haemorrhoidal artery ligation (DG-HAL) and transanal haemorrhoidal dearterialisation (THD).

Technique

With the patient in gynaecological position, local anaesthesia is performed with perianal infiltration in the four quadrants. A dedicated fenestrated anoscope which, at its tip, holds an 8.2 Mhz Doppler microprobe is inserted. The function of this microprobe is to allow identification of the terminal branches of the superior haemorrhoidal arteries. The fenestration on the anoscope, situated just below the Doppler microprobe, allows

putting a stitch (rounded needle and absorbable material) and thus tying the arterial branch found with the Doppler signal. Each arterial branch is ligated about 2–3 cm above the dentate line. The disappearance of Doppler signal documents the correct execution of the manoeuvre. Ligation of terminal branches of the superior haemorrhoidal artery determines a reduction of blood pressure inside the plexuses with subsequent reduction of bleeding and swelling of the haemorrhoidal tissue. In general, the procedure ends after performing 6 ligatures at 11, 12, 2, 5, 7 and 9 o'clock; duration is about 20–30 min.

Results

The first encouraging results were published by Morinaga et al. [73] himself, who reports that 1 month after the procedure of the 116 patients operated, 96 % did not have bleedings anymore and 95 % did not feel any pain; and in 78 % of cases, he reported an improvement of prolapse-related symptoms.

Between 2004 and 2007 also Scheyer et al. [74], Dal Monte et al. [75] and Lienert and Ulrich [76] reported very positive results in their studies: the technique is considered to be well tolerated by patients and relatively painless and can be performed in an outpatient setting under local anaesthesia/sedation. Moreover, in a randomised study in which he compares the technique in question to conventional haemorrhoidectomy, Bursics et al. [77] finds that, although the two techniques are equally efficient, arterial ligation is less painful and allows a quicker return to social life.

It has to be said, however, that in the quoted studies, the majority of patients treated by arterial ligation were affected by second- to third-degree haemorrhoids. In fact, although this technique seems able to provide satisfactory results in haemorrhoids mostly affected by bleeding, similar results are not achievable for haemorrhoids with symptoms of prolapse. The aforementioned Scheyer et al. [74] himself remarks how 60 % of patients with fourth-degree haemorrhoids complained of a residual prolapse and how, on the other hand, only 6.7 % of patients with second-degree haemorrhoids presented with a similar affliction in the postoperative period. The problem seems related to the impossibility of achieving a satisfactory correction of the prolapse in the advanced stages of the disease.

With the aim of overcoming this limit, Morinaga's technique was modified at the end of 2005 with the addition of a "rectoanal-plasty": rectoanal repair (RAR) – hence, the acronym HAL-RAR. This procedure adds to the simple HAL a plication of the redundant haemorrhoidal tissue that is suspended and fastened to the rectal walls as an effect of scar formation. This technical precaution leads to a correction of prolapse-related symptoms (mucorrhea, pruritus, occasional soiling) frequently seen in patients with third- and fourth-degree haemorrhoids. Middleton et al. [78] reports that the addition of this further technical step improves outcome and does not translate into greater pain to the patient.

Comment on Haemorrhoidal Artery Ligation

This method initially proposed by Morinaga has been the source of much perplexity. The therapeutic foundation of the method is that bleeding and haemorrhoidal prolapse are caused by an arterial hyperafflux. Therefore, a reduction in blood flow would cure this. As mentioned before, blood hyperflow to haemorrhoidal cushions is a normal anatomical and physiological condition that allows haemorrhoids to rapidly increase or decrease in size and with that modifies the volume and closure of the anal canal, thus improving continence.

The haematic flow to the cushions is variable and influenced by a number of factors; for example, the middle haemorrhoidal arteries and veins are intrasphincteric, and therefore, blood inflow and outflow are affected by sphincter tone. So blood afflux is variable and cannot be considered an absolute and measurable rate.

The diagnostic instruments currently available are not sensitive enough to detect a difference in blood flow in the haemorrhoidal arteries of two different study groups. Theoretically, in order to confirm the theory of hyperafflux, we would have

to be able to measure, and differentiate, blood inflow in thousands of patients and, in a prospective trial, check and compare the incidence of haemorrhoidal disease between the normal-flow and the hyperflow group. Even if we did establish that patients with haemorrhoidal disease have an increased haemorrhoidal blood flow, we still would not know whether this is a cause or an effect of the disease. The incidence of haemorrhoidal disease is so high that assuming such an odd and rare alteration would be among the pathogenetic factors is at least highly improbable.

Therefore, I think that some of the papers appearing in the literature [79] should be considered unreliable. It is biological nonsense arguing that the supporting tissue of the anal mucosa and haemorrhoids that consists of smooth muscle and elastic fibres, when hypervascularised and hence hyperoxygenated, transforms into frail fibrotic fibres that in turn break and then cause prolapse. It would mean jettisoning what has been confirmed in histochemical findings, i.e. that it is a deficit in blood flow that transforms muscle fibres into fibrotic tissue. It is even more difficult to imagine how haemorrhoids are pulled back up into the anal canal when blood flow is reduced.

However, despite the aforementioned reservations on the therapeutic principles of haemorrhoidal dearterialisation, I have had chance to establish that this method can be efficient in some patients with modest second-degree prolapse, although with a high incidence of recurrences and postoperative internal and external haemorrhoidal thrombosis. From the beginning it has always been my conviction that the efficacy of this method was due to the numerous stitches applied at full thickness to the rectum. These stitches fix the rectal mucosal prolapse, thus keeping it from sliding into the anal canal. Therefore, whether you close the arteries (or not) does not have any influence on the outcome, nor does using the Doppler ultrasound.

Confirmation of these assumptions has nonetheless arrived from the owners of the THD patent. The two surgeons at first had only performed simple ultrasound-guided ligation, but when they determined the procedure's scarce efficacy on the prolapse, they associated a continuous suture to fix haemorrhoids. Later they showed that when performing only the fixing suture, the results were the same. Consequently they abandoned the Doppler.

I know of similar studies that were sent to renowned coloproctology journals and, for inexplicable reasons, have always been rejected.

Operative Treatment

Open Haemorrhoidectomy (Milligan-Morgan)

The so-called "open" haemorrhoidectomy, the first description of which dates back to as long ago as 1937, is still frequently performed in the United Kingdom. Although the operation is based on the procedure originally proposed by Milligan only, it is known always and anyway as Milligan-Morgan haemorrhoidectomy [80].

Technique

The procedure can be performed under general or subarachnoid anaesthesia or by pudendal nerve blockade associated with intravenous sedation. The patient is positioned in lithotomy with the buttocks protruding beyond the edge of the operative table and spread apart. Some surgeons find it useful to infiltrate the perineal region with saline solution to which adrenaline is added, with the double advantage of reducing bleeding and facilitating dissection of the haemorrhoidal pedicles.

For each of the three main haemorrhoidal piles, the overlying skin is grasped with a robust forceps and is retracted outwards. This manoeuvre allows exposure of the inferior margin of the mucosa covering the haemorrhoidal pile. The latter is grasped with another forceps so that three pairs of forceps can be seen in the operative field, one for each haemorrhoidal plexus.

Usually, the first haemorrhoidal pile to be treated is the left lateral one. By holding with the left hand both forceps, that – as already said – grasp skin and mucosa of that pile, and by pulling them together towards the centre of the

anus, the surgeon performs a "V"-shaped incision with the scissors in his right hand. The two margins of this incision run from the mucocutaneous junction to the two sides of the pile and meet at the apex of the "V", about 2–3 cm from the dentate line. He then proceeds cautiously to dissect the haemorrhoidal plexus from the underlying muscle, being careful to preserve the internal sphincter fibres. Progressively, while dissection proceeds upwards, the mucosa is sectioned on both sides with incisions that converge towards the pedicle. This is extremely important in order to leave wide mucosal bridges behind at the end of the procedure. When a pedicle is well isolated, it is transfixed and tied. The haemorrhoidal tissue is then sectioned from its pedicle and removed. The procedure is repeated exactly with the same modalities for the other haemorrhoidal piles. At the end of the operation, three open wounds are visible, similar in shape to three pears whose stalks converge towards the anus. The region is dressed with paraffin-soaked gauze positioned in the anal canal.

Other versions of the operative technique just described are haemorrhoidectomy with LigaSure and laser haemorrhoidectomy.

Haemorrhoidectomy with LigaSure

The technical procedure is extremely simple. In this way to perform an haemorrhoidectomy, haemorrhoidal plexuses are excised by applying electrothermal bipolar energy is delivered by a forceps device. The first results were published by Jayne et al. [81], Palazzo [82], Chung and Wu [83] and by Franklin et al. [84]. One of these publications, by Palazzo, compare this technique with haemorrhoidal dissection by diathermy and with open haemorrhoidectomy, respectively. The author find advantages in terms of operative time, post-operative pain, need for medications and return to social and work activities. The cost of the instrument, however, needs to be considered.

Laser Haemorrhoidectomy

Among the various types of lasers with the widest applications in medicine, carbon dioxide laser and neodymium-based laser (neodymium:yttrium-aluminium garnet, Nd-YAG) proved the most adequate types for surgical proctology. This is due to the specific wavelength of these types of laser that, when in contact with venous tissue, have shown a good coagulation potential with scarce collateral tissue damage. Apart from the obvious set of special instruments, the use of a laser device requires a specific preparation of the medical and paramedical staff.

Having said this, the technique itself is relatively easy: after local anaesthesia is induced, with the patient in prone position and with the aid of a Hill-Ferguson anal retractor, the laser beam is aimed directly at the haemorrhoidal plexus.

The laser light, applied with a dedicated handle, is slowly moved onto the area to be treated until total tissue destruction becomes evident by the appearance of a uniform whitish membrane. The entire procedure requires about 30 min.

Nicholson et al. [85] report that postoperative results regarding early and late complications, need for pain killers and wound healing time are not different from those reported after conventional haemorrhoidectomy.

Wang and colleagues are not of the same opinion [86]. In 1991, they compared haemorrhoidectomy with neodymium laser to closed haemorrhoidectomy (Ferguson technique) in a randomised trial. From their results it emerges that patients after laser haemorrhoidectomy needed less analgesics and had less urinary retention, and the operative time was shorter.

The opinion held by Senagore et al. [87] is different as emerges from one of his publications in 1993. In this randomised trial the author compares haemorrhoidectomy with neodymium laser to the one performed with a "cold" scalpel. Both the procedures were performed by standard closed Ferguson haemorrhoidectomy: there were no statistically significant differences between the two procedures, although a higher incidence of suture dehiscence was recorded in the group of patients treated by laser. Therefore, Senagore concluded that, also because of the higher costs, the use of laser haemorrhoidectomy was not justified.

Closed Haemorrhoidectomy (Ferguson Procedure)

This surgical technique developed by Ferguson and Heaton in 1959 (Rosa) was conceived with the aim of overcoming the common disadvantages attributed to open haemorrhoidectomy [88]. The authors had the aim of removing most of the haemorrhoidal tissue while sparing the anoderm, reducing postoperative serous secretions and preventing stenosis following healing by second intention. To date, this is still the haemorrhoidectomy technique most frequently used in the United States.

Technique

Closed haemorrhoidectomy can be performed under any type of anaesthesia. In case of local anaesthesia, the drug of choice is bupivacaine with adrenaline. Regional anaesthesia with caudal block and subarachnoid anaesthesia are also among the possible options.

Although the procedure is performed by some surgeons with the patient in gynaecological position and although Ferguson himself, in describing his original technique, suggested to position the patient in Sims position, the genupectoral position (jackknife) is currently the one which is most widely used. The buttocks are spread apart with adhesive tape to better expose the anal region. The area is disinfected with iodine solution, and there is no need for hair removal. The anal canal is explored with a Pratt bivalve anoscope in order to allow the surgeon to evaluate in which and in how many quadrants haemorrhoidectomy is necessary. In fact, although the three-quadrant haemorrhoidectomy including the anterior, right posterior and left lateral regions is the most common one, it is not the rule at all. Generally, after exploration with the Pratt anoscope, a Fansler operative anoscope will be inserted; thanks to a fenestration running over the whole length of the anoscope, this device allows avoiding an excessive excision of the anal epithelium, helping to prevent stenosis. The anoscope is positioned in line with the haemorrhoidal plexus to be resected.

The haemorrhoidal plexus and relative anal tag which may be adjacent to it are grasped with a forceps and drawn upwards and towards the centre of the anal canal. With Metzenbaum scissors the tissue below the forceps is incised, from the skin towards the centre of the anus, paying attention to keep the internal sphincter fibres away. Usually the most voluminous haemorrhoidal plexus is excised first. After separating the haemorrhoidal plexus from the internal sphincter over its whole extension, the surgeon proceeds to the ligation/section or to the diathermy coagulation of the vascular pedicle. An absorbable suture is used to close the surgical wound completely.

In order to keep the risk of stenosis as low as possible, the surgeon generally performs wound closure with the anoscope in situ.

Submucosal Haemorrhoidectomy (Parks Procedure)

Parks [89] describes this procedure in 1956. It is substantially an excision of haemorrhoidal tissue performed after incision of the mucosa inside the anal canal and the rectum, followed by mucosal suture: the aim is to reduce healing time and the rate of stenosis.

Technique

Any anal retractor can be used. Also for this procedure, it can be useful to infiltrate the area to be treated with a solution containing adrenaline. The incisions are performed starting from the skin just outside the anal verge, and exerting traction on the skin just beyond the external margin of the wound, the incision is then extended upwards including the rectal mucosa above the haemorrhoid up to 4 cm from the mucocutaneous junction. The haemorrhoidal tissue is exposed and separated from the mucosal folds and from the underlying muscular plane and then removed after being transfixed and tied in proximity of the vascular pedicle. The mucosal folds within the anal canal are reapproximated. The margins of the anoderm below and the rectal mucosa above next to the pedicle ligatures are left open.

Circular Haemorrhoidectomy (Whitehead Procedure)

Described for the first time by Whitehead 130 years ago [90], circular haemorrhoidectomy was initially met with keen interest mainly in the United Kingdom but has now been abandoned almost completely, mostly because of its difficult technical execution.

Technique

The procedure originally described by the author implies a circular incision at the level of the dentate line for each plexus to treat. Through this incision, the surgeon proceeds, moving upwards, to the dissection of the haemorrhoidal tissue. After completing ligation of the vascular pedicle, the haemorrhoidal plexus is removed, and the surgeon proceeds to suture the mucosa to the anal canal at the level of the dentate line.

In a wrong interpretation of the descriptions given by Whitehead, many surgeons sutured the mucosa to the skin at the external anal margin. Apart from leading to frequent dehiscences of the sutures caused by excessive tension, this technical error often resulted in a deformity of the anus characterised by the protrusion of the anal mucosa out of the external anal margin. This complication, consisting of a mucosal ectropion, has become known as "Whitehead deformity", making the illustrious colleague more notorious for a complication for which he was not responsible than for the procedure first described by him.

Results

Whenever the technique is performed following the original indications given by Whitehead the reported results are similar to those of other haemorrhoidectomies. Bonello [91], Wolff and Culp [92], Barrios and Khubchandani [93] agree with this.

Comment on Haemorrhoidectomy

The purpose of surgical haemorrhoidectomy has always been that of removing haemorrhoids as radically as possible, with the minimum possible postoperative pain and limited complications. The Whitehead procedure certainly is the one that allows the most complete excision of haemorrhoidal tissue, but it has never been well accepted because of the anal deformity of the same name. In reality, this is an iatrogenic mucosal ectropion due to the suture placed between the anal mucosa and the dentate line or, by mistake, the anal skin. An almost constant complication of this procedure is anal incontinence, varying from moist anus to faecal incontinence. For those who have no direct experience of the Whitehead procedure, it is sufficient to remember that these sequelae are comparable to those of ultra-low coloanal anastomosis. These complications give us the chance to comment on something I suggest we should keep in mind: removal of haemorrhoids and of anal mucosa implies that the anal canal becomes lined with rectal mucosa; rectal mucosa directly secretes mucus outwards, causing soiling; the rectal contents can no longer be adequately discerned by the receptors of the anal mucosa, and the haemorrhoidal cushions can no longer play their role in improving the closure of the anus so that continence ends up being weakened.

Two banal conclusions can be drawn from the considerations above: in order to adequately perform a retaining function, the muscular anal canal must be covered with anal skin and mucosa, while the haemorrhoids improve this function. In grade IV haemorrhoidal prolapse, the anal canal is covered, entirely or in part, by rectal mucosa prolapsed into the anus which frequently causes soiling. Soiling consists of mucus with a high bacterial load, variable PH and a number of dissolved substances. For this reason mucus can cause bacterial, chemical and also fungal dermatitis. As soiling and dermatitis are caused by haemorrhoidal prolapse, they should be considered correlated symptoms. Hence, surgical treatment of haemorrhoidal disease should also aim at healing soiling and pruritus, and the efficiency of a technique should also be evaluated in relation to the resolution of these symptoms. Instead, in the literature these symptoms are rarely taken into consideration pre- and postoperatively.

Much confusion clearly exists about haemorrhoidal disease when it comes to determining its specific symptoms.

All the longitudinal haemorrhoidectomies – Milligan-Morgan, Fergusson, etc. – imply the removal of longitudinal folds of anal mucosa covering the haemorrhoids. These mucosal folds are the ones that mainly allow the anus to dilate, because they are not directly adherent to the sphincters. Therefore, haemorrhoidectomy inevitably entails a reduction of anal dilatability.

In a certain percentage of cases, which varies considerably in the literature, this deficit is pathological. The tight anal stenoses are due to the almost complete excision of the anal skin and mucosa. The procedures for rotation and sliding of perineal cutaneous flaps restore the anal lumen more or less satisfactorily. But because this type of tissue is not specialised in sensory discrimination and these flaps have no receptors connected to the anal sphincters, the continence function, including the anal opening and closing reflex, results severely and definitely compromised and so is the patients' quality of life.

Severe anal stenosis is reported in the literature at very variable rates. It certainly is an uncommon complication, although there is a feeling that this problem is underestimated. In fact if every proctologist compared the cases of stenosis (obviously occurring in patients operated on elsewhere!) that he had to manage with the overall number of haemorrhoidectomies performed, he would come to the conclusion that his data are not consistent with data reported in the literature.

The historical problem with haemorrhoidectomies is postoperative pain and the long healing time. A very high proportion of patients refuse haemorrhoidectomy even though they have significant symptoms because they know that postoperative recovery is extremely painful. And there is no doubt that this is the popular opinion about this procedure. This leads patients to try whichever remedy in the hope of avoiding an operation. Therefore, tons of rectal ointments and drugs are being used all over the world every day. The result of this is an incalculable waste of money, with frequent iatrogenic damages which are often worse than the disease itself. Very probably this widespread opinion on haemorrhoidectomy has not changed substantially despite our efforts to reassure patients and allay their fears. Certainly postoperative recovery has become significantly more tolerable. It still remains to be clarified whether this is owed to more effective antalgic therapies or to the innovative techniques and technologies proposed. In order to understand and control post-haemorrhoidectomy pain, it is imperative to understand what postoperative incidents are causing it and if these traumas can be modified by adopting different forms of energy for tissue dissection.

Understanding the origin of post-haemorrhoidectomy pain is not easy at all, and there is a risk of being sidetracked by studies that in the last decades have created many expectations and just as many disillusions. The ordinary post-haemorrhoidectomy wound is made up of a muscular base consisting of corrugator ani and smooth sphincter and of two severed margins consisting of anoderm, mucosa and submucosa. The smooth sphincter contains only pressure receptors and is not traversed by sensory fibres directed to the anal mucosa, as these come from the rectum. This means that the only pain sensation perceivable by the smooth sphincter is either caused by specific stimulation of the pressure receptors or by an aspecific stimulus to the sectioned and exposed nerve terminations. The same considerations apply to the mucosa of the anal canal which has few pain and temperature receptors but is endowed with pressure receptors. In fact the only form of pain perceivable in the anal mucosa and haemorrhoids is tensive pain caused by tissue distension, as, for example, in case of thrombosis, oedema or strangulation following RBL. This is confirmed by the fact that any procedure performed above the anoderm is almost painless. Thus the site of post-haemorrhoidectomy pain is in all likelihood the dermal-anodermal region. This region with its very dense somatic innervation contains numerous receptors specialised towards both physical stimuli, like temperature, stretching and pressure, and chemical stimuli, mainly PH.

The idea that the energy adopted for tissue excision (monopolar diathermy, laser, Ultracision, LigaSure) can modify postoperative pain has been and is still being advocated with much emphasis. This conviction can be rebutted with a few obvious considerations. If the use of a type of energy that develops little heat improves pain, it would be ideal to incise and excise the anal skin with a cold scalpel blade to avoid heat-related tissue damage. Clinical experience tells us otherwise. The thermal receptors are stimulated by temperatures different from those typical of the body, both hot and cold, and only when this difference becomes remarkable, the stimulus is perceived and transmitted as painful. There is no doubt that, whichever temperature the tissue reaches during the procedure, the patient does not perceive pain thanks to anaesthesia; after a few minutes, this tissue resumes normal body temperature and so the patient cannot feel pain generated from heat.

It could be objected that there is also significant biological damage to the residual anoderm and the perianal skin, but here too the consideration that the use of a cold blade, with selective coagulation or ligation of the vessels, should cause less damage and less pain, is valid. It should be remembered that tissue repair, both by first intention (closed haemorrhoidectomies) and by second intention (open haemorrhoidectomy) happens thanks to fibrin, fibroblasts and other repair factors exuding from arterial, venous and lymphatic capillaries and deposited on the injured surfaces. This might cause a delay in scarring, and I do think that this is not an unfounded assumption. This adverse event, which has been described in closed haemorrhoidectomies (references), strangely is not reported in the case of Milligan-Morgan procedures.

Experience and data from the literature confirm that the type of energy used do not influence pain. For many years countless papers – whether entirely objective or not – have been reporting significantly better results with the use of a laser. These results have since been irrefutable disproved by subsequent more reliable studies.

I am convinced that all the other forms of energy currently proposed for haemorrhoidectomy will be facing the same fate as laser.

Unitary Theory of Rectoanal Prolapse

In order to introduce the principles of stapled haemorrhoidopexy (SH) and stapled transanal rectal resection (STARR), I think it is useful to briefly present the considerations, clinical observations and original studies which form the rational basis of these techniques.

At the beginning of the 1990s, after performing hundreds of Milligan and Morgan, Ferguson, Whitehead and other procedures, I realised that the postoperative period was a very painful experience for patients and that the sad notoriety of these procedures induced many persons to exclude surgery.

Initially I developed an inferiority complex, thinking I was not able to perform the technique correctly, but later on, after having visited renowned coloproctology centres, I understood that I was obtaining the same results they did and that there was – and is even more so today – an enormous discrepancy between clinical reality and publication of results. I became convinced that a haemorrhoidectomy, whether closed, open, semi-closed or other, did not represent an adequate therapeutic response to the problem. Through the simple analysis of and extensive reflections on the literature, I sensed that the very essence of haemorrhoidal pathology had still not been captured. I came across some incoherences and contradictions that represented a stimulus for me to study of the problem more in depth. Thomson [1] thought that the disruption of the haemorrhoids' supportive tissue caused prolapse. Haas et al. [2], in 1984, demonstrated that haemorrhoidal supportive tissue disintegrates in all subjects after the age of 30, but not everyone has symptomatic haemorrhoids or prolapse.

This information matches the clinical observation that haemorrhoidal prolapse is always associated to rectal mucosal prolapse, that can protrude into the anal canal and then outside the anus. The external prolapse of rectal mucosa is permanent in the so-called fourth-degree prolapse, and so it is clinically comparable to an ectropion.

Non-reducible rectal mucosa prolapse is thus certainly the cause of soiling which in turn causes

perianal dermatitis and hence pruritus. Where and how does rectal mucosa, which prolapses together with the haemorrhoids, return to within the rectum at the end of evacuation in the case of second- and third-degree prolapses?

To answer this question, we performed a defecography in all patients with haemorrhoidal prolapse of grades II, III and IV in addition to routine proctoscopy. The result was that all patients with any degree of haemorrhoidal prolapse presented a rectoanal invagination whose size could not be correlated with the size and grade of the external prolapse.

Moreover, in women a rectocele was almost always associated to rectoanal invagination. Rectoanal invagination and rectocele associated to haemorrhoidal prolapse explain the previously unclear correlation between haemorrhoidal disease, straining and obstructed defecation (OD). If we consider the consistent association of haemorrhoidal prolapse and rectal prolapse, we can exclude that this is just an occasional concomitance of two distinct pathologies as has always have been thought and written. Clearly this must be a clinical and pathological picture of its own. What still had to be determined was a cause-effect relationship: is it the haemorrhoids that, when prolapsed, draw the rectal mucosa down, or is it the rectal prolapse that pushes the haemorrhoids out of the anus? There was no doubt for us that the second hypothesis had to be correct. In fact it is impossible to find a haemorrhoidal prolapse without rectal prolapse, while the opposite is frequently observed. We came up with the theory that rectoanal invagination causes a kink in the superior haemorrhoidal veins which hampers haemorrhoidal venous outflow and leads to dilatation of the haemorrhoids. This dilatation and stretching of the haemorrhoids can cause increased friction and mechanical trauma to the overlying mucosa during the passage of faeces, with subsequent de-epithelisation and bleeding. We think that this clinical condition constitutes what it is defined rather fuzzily as first-degree haemorrhoidal prolapse.

Subsequently rectal invagination extends to the anal canal during evacuation, causes its obstruction and induces increased straining. It is easy to imagine but also demonstrable in dynamic cinedefecography how the faecal bolus pushes both rectal prolapse and haemorrhoids out of the anal canal with force. In fact, only once the prolapse is expelled, the anal canal is cleared and evacuation can start.

The above-mentioned clinical studies have led us to a revolutionary conclusion that is in conflict with all the traditional ideas on the pathogenesis of haemorrhoidal disease: haemorrhoidal prolapse and all its related symptoms constitute a pathology secondary to the internal rectal prolapse; the rupture of the supportive tissue of the haemorrhoids is a necessary, but not sufficient, precondition for a prolapse to occur. In fact, rupture of supporting fibres is a physiological phenomenon typical of aging (Haas [2]) and it does not necessarily imply haemorrhoidal prolapse. Seemingly, in young subjects the rupture of the Treitz fibres is caused when the rectal prolapse repeatedly pushes against the haemorrhoidal cushions. Haemorrhoidal prolapse is therefore only one of a number of possible clinical manifestations of an internal rectal prolapse.

This new theory explains the correlation between haemorrhoidal disease and obstructed defecation. In fact, rectoanal invagination is also the main cause of the obstructed defecation syndrome.

The limited scope of this chapter does not allow us to go into detail about all the clinical aspects related to rectoanal prolapse and the studies that led us to some conclusions. It is however necessary to give at least a short summary in order to explain the rational basis of the techniques for the therapy of haemorrhoidal prolapse that we are about to describe. Internal rectal prolapse, whether associated or not to anal, mucosal and haemorrhoidal prolapse, can be a mucosal rectal prolapse (about 10 % of our cases) or a full-thickness rectal prolapse.

When performing a baseline 2-view X-ray of the empty rectum with barium contrast, we can see that some patients present with a rectum the shape of which, especially distally, indicates a detachment from the sacral-coccygeal plane; this type of rectum is usually folded on itself, with unnatural loops lying on the perineum, and is longer than usual. We have called this conformation “rectal redundancy”.

When performing a varied dynamic rectal videodefecography in patients with rectal redundancy, i.e. with only the sigmoid filled with barium and potato starch, we note that such a redundancy always causes an obstacle to the transit of barium into the rectal lumen, causing more intense straining and repeated attempts at evacuating.

During straining it is possible to observe how this rectal redundancy can assume different morphological aspects including various combinations: simple or multiple invagination, rectocele caused by rectal dilatation or by formation of a loop and partial or total outward expulsion of the rectum.

In patients with haemorrhoidal prolapse, a good impregnation of the anal canal allows to visualise how a rectoanal invagination pushes haemorrhoids and anal mucosa outside. It is also interesting to observe that in some patients a descent of the Douglas pouch or formation of an enterocele can be seen during straining; by compressing the rectum from above and pushing it towards the sacrum, this facilitates emptying of the rectal contents.

Enterocele and Douglas pouch dislocation are always associated to an excessive perineum lowering. These pelvic alterations can disappear completely or partially and can persist at the end of straining. With regard to their behaviour, we have divided these pelvic alterations into stable and dynamic ones. They are indeed caused by excessive straining and in our opinion should be considered supporting mechanisms compensating for the incapacity of a prolapsed rectum to empty physiologically.

With regard to the nature of the rectocele, I would like to point out that cadaveric studies and ultrasonographic mapping of the rectum in patients with a similar clinical and defecographic picture have clearly demonstrated that the only perceivable alteration is the thinning or disappearance of the muscular layer of the rectum. This type of defect begins just above the anal canal and extends variably upwards. As the rectal ampulla is no longer effectively supported by the muscular layer, it can expand anteriorly and, after occupying the perineal body, push onto the posterior vaginal wall, causing a colpocele. Large rectoceles can dilate the vaginal wall abnormally causing secondary structural damage. In any case, a rupture of the famous rectovaginal septum – the definition, function and existence of which have always been controversial – cannot be considered a primary cause of rectocele. In numerous cadaveric dissections, we have actually never been able to detect such a septum, and we therefore think that this is probably a wrong definition given by gynaecologists (out of self-interest?). We have recently received a clear confirmation of this theory by pathologists who never detected such a "septum" in specimens sent in by gynaecologists. We thought that these – unfortunately rather detailed – preliminary remarks were necessary to make the following conclusion understandable: haemorrhoidal prolapse is a pathology secondary to and consequent upon rectal prolapse, be it only mucosal or full thickness.

We therefore consider the clinical and pathological distinction between haemorrhoidal disease, rectal prolapse and rectocele an artificial one. Although the haemorrhoidal prolapse is the pathological alteration that causes the typical symptoms of haemorrhoidal disease, it must be considered simply an external manifestation of a prolapse of – initially – the rectum that subsequently and progressively can cause a prolapse of the anal mucosa, the haemorrhoidal cushions and the anoderm. Not always does an internal rectal prolapse cause a mucous and haemorrhoidal prolapse associated to it, but all haemorrhoidal prolapses are invariably associated to rectal prolapse. For this reason a more correct definition would be that of a rectoanal prolapse which would provide a more correct description of the anatomical and pathological condition. An in-depth revision of these pathologies and a clinical reclassification based on a new theoretical foundation is therefore necessary. We have proposed a single combined classification of these pathologies termed "unitary theory of rectoanal prolapse".

Based on the results of our studies and our observations, we came to the conclusion that a treatment consisting of the correction of internal rectal prolapse could also represent a rational treatment for haemorrhoidal prolapse, as it would

cure the cause that determines it. Moreover, the resection of the internal rectal prolapse would also resolve obstructed defecation if present, both because it would eliminate the mechanical obstacle and because resection of the distal rectum would include removing any rectoceles present, leading to improved of rectal compliance. Given the focus of this chapter, we will limit ourselves to stressing the fact that bleeding, thrombosis and haemorrhoidal oedema are only a few of the possible symptoms of the prolapses defined as recto-anal, and therefore obstructed defecation and continence disorders have to be taken into consideration when taking the history of these patients.

The rectal prolapse associated to a haemorrhoidal prolapse can present with different sizes, and there is no correlation in terms of size between haemorrhoidal and rectal prolapse. For this reason the simple clinical evaluation of an external haemorrhoidal prolapse is not predictive of the size of the rectal prolapse that has to be removed, and so it does not allow us to determine the technique that needs to be chosen. Histologically, a simple mucosal prolapse is a detachment of mucosa and submucosa from the muscular layer of the rectum, and given their increased length, it presents as a redundancy. Full-thickness prolapse is generally characterised by a lengthening of the rectum because of structural alterations of the muscular layers. The rectum tends to form multiple loops that fold on themselves. In other cases rectal prolapse can be due to slippage of the whole rectum-sigmoid. In this case the natural evolution is a complete external rectal prolapse. Mucous rectal prolapses can be resected with the stapled anopexy technique. STARR is reserved to large mucosal prolapses or to full-thickness prolapses. STARR can be performed by means of two PPH devices or with the more recently introduced curved stapler that goes by the (unfortunate) name of TRANSTAR.

Stapled Haemorrhoidopexy (SH)

Technique

Also known as the PPH procedure, Longo procedure, stapled anopexy and circumferential mucosectomy, PPH is a technique developed in 1993 that reduces the prolapse of haemorrhoidal tissue by excising a doughnut-like ring of the prolapsed rectal mucosa with a circular stapling device: with this the haemorrhoidal cushions, anal mucosa and anoderma are lifted and permanently fixed in their anatomical position [94], and a haemorrhoidal prolapse during defecation is prevented.

The procedure can be performed under subarachnoid anaesthesia; the patient is placed in lithotomic position. A PPH-01 or PPH-03 kit (Ethicon Endo-Surgery, Cincinnati, Ohio) is necessary. The introduction of the circular anal dilator (CAD) causes the reduction of the anal prolapse into the rectum. After removing the obturator, the prolapsed rectal mucosa falls into the lumen of the dilator. The purse-string anoscope (PSA) is then introduced through the dilator. This anoscope will push the prolapsing mucosa back against the rectal wall along a 270° circumference, while the mucous membrane that protrudes through the anoscope window can be easily captured with a stitch (Prolene TM 00, Ethicon). By rotating the anoscope, it will be possible to complete a purse-string suture around the entire rectal circumference, 2–3 cm above the haemorrhoidal apex.

A PPH-01/PPH-03 stapler is opened to its maximum position. Its head is introduced until crossing the purse-string which is then tied with a knot. The ends of the suture are knotted externally. Then the stapler is partially tightened while keeping the casing outside. Once half the casing is inserted into the CAD, it is pushed against the purse-string, and while exerting moderate traction on the ends of the suture, the instrument is tightened. Keeping the stapling device in the maximum closed position, for approximately 30 s, may improve the haemostasis. Firing the stapler releases a double staggered row of titanium staples through the tissue. The circular stapler knife excises the tissue. A circumferential column of mucosa is removed. Finally, the staple line is examined using the anoscope. Additional haemostasis can be achieved by stitches (Vicryl TM 2-0, Ethicon).

Results

A recent systematic review [95] has allowed us to document the fact that a huge number of scientific publications are available in the literature about the PPH procedure: there are 29 publications [96–124] on 25 randomised clinical trials comparing PPH stapled haemorrhoidopexy with conventional haemorrhoidectomy. They included a total of 1,918 patients, of whom 971 underwent stapled haemorrhoidopexy (PPH procedure) and 947 had surgical haemorrhoidectomy. The main results are reported here.

Procedure Time

In the 23 trials [96, 98, 100, 102, 103, 105–124] in which it was possible to calculate, it was found that the PPH procedure stood out for its significantly shorter operating time compared with conventional haemorrhoidectomy [mean operating time, 17.55 vs. 28.90 min; weighted mean difference (WMD) – 11.35 min; $P=0.006$].

Pain

The PPH procedure caused significantly less postoperative pain than conventional surgery. Twenty-three trials [96, 98, 100, 102–108, 110–112, 114, 115, 117–124], reported significantly less pain after PPH as evidenced by reduction of the pain scores at rest and on defecation by 42.3 %.

Recovery

There was a faster surgical and functional recovery after stapled haemorrhoidopexy. The PPH haemorrhoidopexy allowed a faster functional recovery with shorter time off work (WMD – 8.45 days; $P<0.00001$) and earlier return to normal activities (WMD – 15.85 days; $P=0.03$).

Patient Satisfaction

Significantly more patients in the PPH than in the conventional haemorrhoidectomy group rated the procedure as satisfactory [93.3 vs. 86.4 %; odds ratio (OR) 2.33; $P=0.003$] [98, 102, 105, 106, 112, 119, 123].

Re-intervention

The PPH procedure did not increase the overall need of surgical (OR, 1.27; $P=0.4$) and nonsurgical (OR, 1.07; $P=0.82$) re-intervention compared with conventional haemorrhoidectomy [96, 98, 100, 103–105, 107–110, 112, 114, 116–119, 121–123].

Bleeding

There was no significant difference in the amount of intraoperative bleeding ($P=0.26$) or the incidence of early postoperative bleeding (bleeding within 24 h of surgery; $P=0.11$). At more than 1 day after surgery, the PPH procedure was associated with significantly less risk of bleeding (9.8 vs. 15.1 %; OR, 0.52 $P=0.001$) [96, 100, 102–106, 108–111, 114–116, 118, 124]. There was no difference between the groups regarding the need for readmission as a result of bleeding (OR, 0.63; $P=0.67$), blood transfusion (OR, 0.64; $P=0.54$), or further nonoperative (OR, 4.06, $P=0.08$) or operative interventions for bleeding (OR, 1.02; $P=0.95$).

Perianal Complications

There was no significant difference between the two procedures regarding early (OR, 1.82; $P=0.52$) [21, 24, 28, 30, 36] or late anal stenosis (OR, 0.69; $P=0.33$) [96, 98, 100, 103, 105, 107, 108, 111, 112, 115, 116, 124], anal fissure (OR, 0.93; $P=0.88$) [96, 105, 112, 114, 116, 117, 123, 124] or perianal fistula (OR, 0.25; $P=0.23$) [107, 110, 112, 116, 117].

Early Recurrence

There was no significant difference between the two groups with regard to early postoperative recurrence (within 6 months) or persistence of symptoms from haemorrhoids: 24.8 and 31.7 % after PPH procedure and conventional haemorrhoidectomy, respectively (OR, 0.68; $P=0.08$) [98, 108, 111, 114, 117, 122, 124]. There was no difference in the need for further operation for early recurrent haemorrhoids (OR, 0.71; $P=0.69$).

Late Recurrence

The incidence of recurrent haemorrhoids at 1 year or more after surgery was higher after stapled haemorrhoidopexy (5.7 vs. 1 %; OR, 3.48, $P=0.02$). However, the overall incidence of

recurrent or persistent symptoms from haemorrhoids was similar in the groups (PPH vs. conventional: 25.3 vs. 18.7 %; OR, 1.57; $P=0.07$) [98, 106–108, 111, 114, 117].

Quality of Life

Three trials [96, 100, 109] addressed the quality of life after surgery. There was no significant difference in quality of life after either surgical procedure, as both the Short-Form 36 Quality of Life questionnaires [97, 109] and the Eypasch Gastrointestinal Quality of Life instrument [100] showed. However, there was a tendency towards higher median physical and mental scores after PPH procedures.

Cost-Effectiveness

Four trials [100, 103, 109, 120] investigated the cost-effectiveness of stapled haemorrhoidopexy compared with conventional surgery. When both the operating cost and hospital stay charges were taken into account, conventional haemorrhoidectomy was more expensive than the PPH procedure, although the differences were not statistically significant [103, 109]. Thus, the cost of the disposable stapler was offset by a shorter hospital stay. In an Asian study [100], where hospital charges are less expensive than in the West, the total medical cost was higher after the PPH procedure (US $1,283.09 ± T 31.59 vs. US $921.17 ± 16.85).

In light of all the above-mentioned considerations, PPH stapled haemorrhoidopexy is safe with many short-term benefits, and long-term results are similar to the conventional procedure.

Stapled Transanal Rectal Resection (STARR)

Technique

STARR was proposed by Longo in 1998. We suggest performing the procedure under subarachnoid anaesthesia with the patient in lithotomic position.

Two PPH-01 or PPH-03 kits (Ethicon Endo-Surgery, Cincinnati, Ohio) are necessary. The anal canal is checked digitally, in order to ensure optimal relaxation. Any haemorrhoidal prolapse or procidentia is reduced using a gauze pack inserted into the anal canal. Four skin stitches are applied to fix the circular anal dilator (CAD) which is lubricated with Vaseline before being inserted. A swab is inserted and slowly retracted to assess the extent of the invagination. A spatula is passed through the posterior window of the CAD to protect the posterior rectal wall. A purse-string suture anoscope (PSA) is used to place 2–3 anterior semicircular sutures (Prolene TM 00, Ethicon) between 9 and 3 o'clock, the most caudal one 2–3 cm above the haemorrhoidal apex, the following ones in 2 cm steps. The homolateral threads are tied under moderate tension. A lubricated maximally opened stapler is introduced with the head passing beyond the sutures. The bundled threads are pulled through the holes of the casing and clamped. Keeping the stapler head in a stable position while the threads are pulled tight, the stapler is partially closed until the resistance of the first suture is felt. By that it is ensured that the edge of the casing is placed above the haemorrhoids and anal ring. Prior to full stapler closure, a vaginal spatula is inserted, and using two fingers the integrity of the vaginal wall is checked. The stapler is closed, fired and removed. Lateral "dog ears" joined by a small bridge which has to be cut are observed frequently. In case of relevant bleeding, haemostatic stitches (Vicryl TM 2-0, Ethicon) are applied. The spatula is now repositioned to protect the anterior rectal wall. The first posterior semicircular suture is placed starting from the base of the left dog ear and moving on to the right dog ear, the second one from the left dog ear's apex to the right one. The posterior prolapse is then resected as described above. Haemostasis is achieved using haemostatic stitches and checked carefully.

Please note: The number of anterior sutures is related to the extension of the invagination. The semicircular sutures on the anterior and posterior wall of the rectum can be substituted by a short running suture. Generally, this option ensures a more homogeneous traction on the rectal walls and, at present, is preferred by colorectal surgeons in Italy.

We suggest to oversew residual dog ears and to apply four to five stitches to reinforce the anterior and posterior staple line. A strip of Vaseline gauze tied to a suture should be introduced to prevent the formation of submucosal haematomas and to facilitate diagnosis of postoperative bleeding. A urinary catheter was inserted in all cases.

Results

Recently, in an attempt to prevent the incidence of failures after SH caused by incomplete resection of the prolapsed tissue (due to the limited volume of the stapler casing), the STARR procedure was adopted successfully for those patients in which a large prolapse was associated with the haemorrhoidal disease. Boccasanta et al. [125] stated that, in patients with an association of prolapsed haemorrhoids and large rectal prolapsed, STARR results in a more complete resection of the prolapsed tissue than SH, with equal morbidity and a significantly lower incidence of residual disease and skin tags. The author used the circular anal dilator, CAD, in order to determine the appropriate surgical technique.

Furthermore, as reported in a recent randomised multicentre trial involving more than 400 patients [126], even if both the PPH-01 and PPH-03 kit can be used, the use of the PPH-03 stapler instead of the PPH-01 ensures a statistically significant reduction of intraoperative bleeding and a significant decrease of operative time.

Comment on SH and STARR

What emerges clearly from a review of the available literature on stapled haemorrhoidopexy and STARR is that many surgeons consider stapled haemorrhoidopexy a procedure indicated for haemorrhoidal prolapse and STARR an operation exclusively indicated in cases of obstructed defecation due to internal rectal prolapse and rectocele. Others consider that in case of haemorrhoidal prolapse, indications for STARR should be limited to patients with associated OD [127]. It is fundamental to revise and adjust these ideas regarding indications for SH and STARR in order to obtain optimal results. As said before, haemorrhoidal prolapse is always a consequence of an internal rectal prolapse of variable size and not correlated to the degree and dimensions of muco-haemorrhoidal prolapse. This implies that a modest external prolapse can be associated to a significant internal prolapse. If we perform SH in these kinds of patients, we will certainly leave a residual internal rectal prolapse behind. We believe that this may predispose for a higher rate of recurrences and may also be the reason why a possibly associated OD is not cured or, worse, even aggravated as the residual prolapse can be jammed inside the anastomosis, especially if a fibrotic ring forms.

It is therefore important to state clearly that for haemorrhoidal prolapses, whether symptoms of obstructed defecation are associated or not, STARR is the procedure of choice whenever an important mucosal prolapse is detected and in all the cases in which there is a full-thickness prolapse or a rectocele. I hope not to shock anyone by confessing that in the last few years, I have myself performed STARR in about 95 % of patients with muco-haemorrhoidal prolapse. Thanks to this decision the rate of recurrences has dropped, at 3 years of follow-up, from 4.9 to 0.4 %. Also with regard to curing OD as one of the complications of this procedure, the results are much more satisfactory. Paradoxically postoperative pain and bleeding have also decreased. In any case, the key aspect is that after so many years the theory has been proven that haemorrhoidal prolapse is secondary to rectal prolapse and that it can be effectively cured by rectal prolapsectomy sparing the haemorrhoids.

Generally, with regard to the advantages of SH and STARR, reduced postoperative pain and faster return to work are frequently highlighted. In my personal opinion the main advantages are resolution of OD (which is often associated), efficient outcome regarding soiling and continence, and the rare incidence of stenosis and, if they occur, the relatively easy treatment of stenosed anastomoses. Now that initial scepticism about SH and STARR as a cure for haemorrhoids has been overcome, the usual detractors insist on a

supposedly higher rate of recurrences following these procedures, which has been proven absolutely incorrect, and on supposed severe complications. Obviously complications can occur, but after three million procedures, only very few cases have been reported and overemphasised with the support of some compliant journal that has published a number of articles without the necessary verifications on the trustworthiness of results. In fact, if some of the articles that report on severe and frequent complications were reliable [128], one would have to suspect a sadomasochistic tendency among thousands of surgeons performing this technique and an inclination to economical failure on the part of the five new companies that have copied the original PPH.

This whole chapter can be summarised by saying that prolapsed and hence symptomatic haemorrhoids are only an epiphenomenon of an internal rectal prolapse which is the primary pathology. Therefore, by adequately treating the internal rectal prolapse, haemorrhoidal disease and all the other symptoms caused by rectoanal prolapse are cured.

References

1. Thomson WHF. The nature of haemorrhoids. Br J Surg. 1975;62:542.
2. Haas PA, Fox Jr TA, Haas GP. The pathogenesis of hemorrhoids. Gut. 1977;18:651.
3. Gass OC, Adams J. Haemorrhoids: aetiology and pathology. Am J Surg. 1950;79:40–3.
4. Goligher J, Duthie H, Nixon H. Surgery of the anus, rectum and colon. London: Balliere Tindall; 1984. p. 5.
5. Andrews E. The treatment of haemorrhoids by injection. Med Rec. 1879;15:451.
6. Edwards FS. The treatment of piles by injection. BMJ. 1888;2:815.
7. Kelsey CB. How to treat haemorrhoids by injection of carbolic acid. N Y Med J. 1885;42:545.
8. Terrell EH. The treatment of haemorrhoids by new method. Trans Am Proctol Soc. 1916;5:65.
9. Anderson HG. The "injection" method for the treatment of haemorrhoids. Practitioner. 1924;113:339.
10. Khoury GA, Lake SP, Lewis MCA, et al. A randomized trial to compare single with multiple phenol injection treatment of haemorrhoids. Br J Surg. 1985;72:741.
11. Dencker H, Hjorth N, Norryd C, et al. Comparison of results with different methods of treatment of internal haemorrhoids. Acta Chir Scand. 1973;139:742.
12. Santos G, Novell JR, Chir M, et al. Long term results of large-dose, single-session phenol injection sclerotherapy for hemorrhoids. Dis Colon Rectum. 1993; 36:958.
13. Ambrose NS, Morris D, Alexander-Williams J, et al. A randomized trial of photocoagulation or injection sclerotherapy for the treatment of first- and second-degree hemorrhoids. Dis Colon Rectum. 1985;28: 238.
14. Gartell PC, Sheridan RJ, McGinn FP. Outpatient treatment of hemorrhoids: a randomized clinical trial to compare rubber band ligation with phenol injection. Br J Surg. 1985;72:478.
15. Rosser C. Chemical rectal stricture. JAMA. 1931;96: 1762.
16. Adami B, Eckardt VF, Suermann RB, et al. Bacteremia after proctoscopy and hemorrhoid injection sclerotherapy. Dis Colon Rectum. 1981;24:373.
17. Ribbans WJ, Radcliffe AG. Retroperitoneal abscess following sclerotherapy for hemorrhoids. Dis Colon Rectum. 1985;28:188.
18. Guy RJ, Seow-Choen F. Septic complications after treatment of haemorrhoids. Br J Surg. 2003;90:147.
19. Blaisdell PC. Prevention of massive hemorrhage secondary to hemorrhoidectomy. Surg Gynecol Obstet. 1985;106:485.
20. Barron J. Office ligation of internal hemorrhoids. Am J Surg. 1963;105:563.
21. Barron J. Office ligation treatment of hemorrhoids. Dis Colon Rectum. 1963;6:109.
22. Corman ML. Hemorrhoids. In: Corman ML, editor. Colon and rectal surgery. Philadelphia: Lippincott Williams & Wilkins; 2005.
23. Mattana C, Maria G, Pescatori M. Rubber band ligation of hemorrhoids and rectal mucosal prolapse in constipated patients. Dis Colon Rectum. 1989;32:372–5.
24. Bartizal J, Slosberg P. An alternative to haemorrhoidectomy. Arch Surg. 1977;112:534.
25. Lee HH, Spencer RJ, Beart Jr RW. Multiple hemorrhoidal bandings in a single session. Dis Colon Rectum. 1994;37:37–41.
26. Walker AJ, Leicester RJ, Nicholls RJ, Mann CV. A prospective study of infrared coagulation, injection and rubber band ligation in the treatment of haemorrhoids. Int J Colorectal Dis. 1990;5:113–6.
27. Marshman D, Huber Jr PJ, Timmerman W, Simonton CT, Odom FC, Kaplan ER. Hemorrhoidal ligation. A review of efficacy. Dis Colon Rectum. 1989;32(5): 369–71.
28. Wrobleski DE, Corman ML, Veidenheimer MC, Coller JA. Long-term evaluation of rubber ring ligation in hemorrhoidal disease. Dis Colon Rectum. 1980;23:478–82.
29. Savioz D, Roche B, Glauser T, Dobrinov A, Ludwig C, Marti MC. Rubber band ligation of hemorrhoids: relapse as a function of time. Int J Colorectal Dis. 1998;13(4):154–6.
30. Bayer I, Myslovaty B, Picovsky BM. Rubber band ligation of hemorrhoids. Convenient and economic treatment. J Clin Gastroenterol. 1996;23:50–2.

31. Armstrong DN. Multiple hemorrhoidal ligation: a prospective randomized trial evaluation a new technique. Dis Colon Rectum. 2003;46:179–86.
32. O'Regan PJ. Disposable device and minimally invasive technique for rubber band ligation of hemorrhoids. Dis Colon Rectum. 1999;42:683–5.
33. Poon GP, Chu KW, Lau WY. Conventional vs. triple rubber band ligand for hemorrhoids. A prospective, randomized trial. Dis Colon Rectum. 1986;29:836–8.
34. Khubchandani IT. A randomized comparison of single and multiple rubber band ligations. Dis Colon Rectum. 1983;26:705–8.
35. Lau WY, Chow HP, Poon GP, Wong SH. Rubber band ligation of three primary hemorrhoids in a single session. A safe and effective procedure. Dis Colon Rectum. 1982;25:336–9.
36. Templeton JL, Spence RA, Kennedy TL, Parks TG, Mackenzie G, Hanna WA. Comparison of infrared coagulation and rubber band ligation for first and second degree haemorrhoids: a randomized prospective clinical trial. Br Med J (Clin Res Ed). 1983;286:1387–9.
37. Ambrose NS, Hares MM, Alexander-Williams J, Keighley MR. Prospective randomized comparison of photocoagulation and rubber band ligation in treatment of haemorrhoids. Br Med J (Clin Res Ed). 1983;286:1389–91.
38. O'Hara VS. Fatal clostridial infection following hemorroidal banding. Dis Colon Rectum. 1980;23:570.
39. Clay III LD, White Jr JJ, Davidson JT, et al. Early recognition and successful management of pelvic cellulitis following hemorrhoidal banding. Dis Colon Rectum. 1986;29:579.
40. Quevado-Bonilla G, Farkas AM, Abcarian H, et al. Septic complication of hemorrhoidal banding. Arch Surg. 1988;123:650.
41. Russell TR, Donohue JH. Hemorrhoidal banding: a warning. Dis Colon Rectum. 1985;28:291.
42. Scarpa FJ, Hills W, Sabetta JR. Pelvic cellulitis: a life-threatening complication of hemorrhoidal banding. Surgery. 1988;103:383.
43. Sim AJW, Murie JA, Mackenzie I. Three-years follow-up study on the treatment of first- and second-degree hemorrhoids by sclerosant injection or rubber band ligation. Surg Gynecol Obstet. 1983;157:534.
44. Wechter DG, Luna GK. An unusual complication of rubber band ligation. Dis Colon Rectum. 1987;30:137.
45. Bat L, Malzer E, Koler M, et al. Complications of rubber band ligation of symptomatic internal hemorrhoids. Dis Colon Rectum. 1993;36:287.
46. Cooper IS, Lee AS. Cryostatic congelation: a system for producing a limited, controlled region of cooling or freezing of biologic tissues. J Nerv Ment Dis. 1961;133:259.
47. Gill W, Fraser J, da Costa J, et al. The cryosurgical lesion. Am Surg. 1970;36:437.
48. Lewis MI. Cryosurgical hemorrhoidectomy: a follow-up report. Dis Colon Rectum. 1972;15:128.
49. White AC. Liquid air: its application in medicine and surgery. Med Rec. 1899;56:109.
50. Williams KL, Haq IU, Elem B. Cryodestruction of haemorrhoids. BMJ. 1973;1:666.
51. Kaufman HD. Haemorrhoids: an 'out-patient package. J R Coll Surg Edinb. 1978;23:40–3.
52. Southam JA. Haemorrhoids treated by cryotherapy: a critical analysis. Ann R Coll Surg Engl. 1983;65:237–9.
53. Berry AR, D'Acosta EFD. The treatment of haemorrhoids by cryosurgery. J R Coll Surg Edinb. 1978;23:37–9.
54. Smith LE, Goodreau JJ, Fouty WJ. Operative hemorrhoidectomy versus cryodestruction. Dis Colon Rectum. 1979;22:10.
55. O'Callaghan JD, Matheson TS, Hall R. Inpatient treatment of prolapsing piles: cryosurgery versus Milligan-Morgan haemorrhoidectomy. Br J Surg. 1982;69:157–9.
56. Goligher JC. Cryosurgery for hemorrhoids. Dis Colon Rectum. 1976;1976:213–8.
57. Keighley RB, Williams NS. Haemorrhoidal disease. In: Keighley RB, Williams NS, editors. Surgery of the anus, rectum and colon. Saunders Co. Lts; 2000.
58. Nath G, Kreitmaier A, Kiefhaber P, et al. Neue Infrarotkoagulationsmethode. Verhaundlungsband des 3 Kongresses der Deutscher Gesellschaft fur Gastroenterologie, Munich. Erlangen: Perimed; 1976.
59. Nieger A. Haemorrhoid in everyday practice. Proc Natl Acad Sci U S A. 1979;2:22.
60. Marques CF, Nahas SC, Nahas CS, et al. Early results of the treatment of internal hemorrhoid disease by infrared coagulation and elastic banding: a prospective randomized cross-over trial. Tech Coloproctol. 2006;10:312–7.
61. Weintein SJ, Rypins EB, Houck J, et al. Single-session treatment for bleeding hemorrhoids. Surg Gynecol Obstet. 1987;165:479.
62. Jensen DM, Jutabha R, Machicado GA, et al. Prospective randomized comparative study of bipolar electrocoagulation versus heater probe for treatment of chronically bleeding internal hemorrhoids. Gastrointest Endosc. 1997;46:435–43.
63. Randall GM, Jensen DM, Machicado GA, et al. Prospective randomized comparative study of bipolar versus direct current electrocoagulation for treatment of bleeding internal hemorrhoids. Gastrointest Endosc. 1994;40:403–10.
64. Hilton CP, Morris DL. A randomized trial comparing direct current therapy and bipolar diathermy in the outpatient treatment of third-degree hemorrhoids. Dis Colon Rectum. 1990;33:931–2.
65. Zinberg SS, Stern DH, Furman DS, Wittles JM. A personal experience in comparing three nonoperative techniques for treating internal hemorrhoids. Am J Gastroenterol. 1989;84:488–92.
66. Varma JS, Chung SC, Li AK. Prospective randomized comparison of current coagulation and injection sclerotherapy for the outpatient treatment of hemorrhoids. Int J Colorectal Dis. 1991;6:42–5.
67. Norman DA, Newton R, Nicholas GV. Direct current electrotherapy of internal hemorrhoids: an effective,

safe and painless out-patient approach. Am J Gastroenterol. 1989;84:482–7.
68. Dennison AR, Whiston RJ, Rooney S, et al. The management of hemorrhoids. Am J Gastroenterol. 1989;84:475.
69. Hinton CP, Morris DL. A randomized trial comparing direct current therapy and bipolar diathermy in the outpatient treatment of third-degree hemorrhoids. Dis Colon Rectum. 1990;22:931.
70. Dennison AR, Whiston RJ, Rooney S, et al. A randomized comparison of infrared photocoagulation with bipolar diathermy for the outpatient treatment of hemorrhoids. Dis Colon Rectum. 1990;33:32.
71. Griffith CDM, Morris DL, Wherry DC, et al. Outpatient treatment of hemorrhoids: a randomized trial comparing contact bipolar diathermy with rubber ring ligation. Coloproctology. 1987;9:332.
72. Yang R, Migikovsky B, Peicher J, et al. Randomized, prospective trial of direct current versus bipolar electrocoagulation for bleeding internal hemorrhoids. Gastrointest Endosc. 1993;39:766.
73. Morinaga K, Hasuda K, Ikeda T. A novel therapy for internal hemorrhoids: ligation of the hemorrhoidal artery with a newly devised instrument (Moricon) in conjunction with a Doppler flow meter. Am J Gastroenterol. 1995;990:610–3.
74. Scheyer M, Antonietti E, Rollinger G, Mall H, Arnold S. Doppler-guided hemorrhoidal artery ligation. Am J Surg. 2006;191:89–93.
75. Dal Monte PP, Tagariello C, Sarago M, Giordano P, Shafi A, Cudazzo E, et al. Transanal haemorrhoidal dearterialisation: nonexcisional surgery for the treatment of haemorrhoidal disease. Tech Coloproctol. 2007;11:333–8.
76. Lienert M, Ulrich B. Doppler-guided ligation of the hemorrhoidal arteries. Report of experiences with 248 patients. Dtsch Med Wochenschr. 2004;129:947–50.
77. Bursics A, Morvay K, Kupcsulik P, Flautner L. Comparison of early and 1-year follow-up results of conventional hemorrhoidectomy and hemorrhoid artery ligation: a randomized study. Int J Colorectal Dis. 2004;19:176–80.
78. Middleton SB, Lovegrove RE, Reece-Smith H. Management of haemorrhoids: symptoms govern treatment. BMJ. 2008;336:461.
79. Ratto C, Donisi L, Parello A, Litta F, Doglietto GB. Evaluation of transanal hemorrhoidal dearterialization as a minimally invasive therapeutic approach to hemorrhoids. Dis Colon Rectum. 2010;53: 803–11.
80. Milligan ETC, Morgan CN, Jones LE, et al. Surgical anatomy of the anal canal, and operative treatment of haemorrhoids. Lancet. 1937;2:1119.
81. Jayne DG, Botterill I, Ambrose NS, et al. Randomized clinical trial of Ligasure versus conventional diathermy for day case haemorrhoidectomy. Br J Surg. 2002;89:428.
82. Palazzo FF, Francis DL, Clifton MA. Randomized clinical trial of Ligasure versus open haemorrhoidectomy. Br J Surg. 2002;89:154–7.
83. Chung YC, Wu HJ. Clinical experience of sutureless closed hemorrhoidectomy with LigaSure. Dis Colon Rectum. 2003;46:87–92.
84. Franklin EJ, Seetharam S, Lowney J, Horgan PG. Randomized, clinical trial of Ligasure vs conventional diathermy in hemorrhoidectomy. Dis Colon Rectum. 2003;46:1380–3.
85. Nicholson JD, Halleran DR, Trivisonno DP, et al. The efficacy of the contact sapphire tip Nd:YAG laser hemorrhoidectomy. Poster presented at the 89th annual meeting of the America Society of Colon and Rectal Surgeons, St. Louis, April 29–May 4, 1990.
86. Wang JY, Chang-Chien CR, Chen J-S, et al. The role of laser in hemorrhoidectomy. Dis Colon Rectum. 1991;34:78.
87. Senagore A, Mazier WP, Luchtefeld MA, et al. Treatment of advanced hemorrhoidal disease: a prospective, randomized comparison of cold scalpel versus contact Nd:YAG laser. Dis Colon Rectum. 1993;36:1042.
88. Ferguson JA, Heaton JR. Closed hemorrhoidectomy. Dis Colon Rectum. 1959;2:176.
89. Parks AG. Surgical treatment of haemorrhoids. Br J Surg. 1956;43:337.
90. Whitehead W. The Surgical treatment of haemorrhoids. BMJ. 1882;1:148.
91. Bonello JC. Who's afraid of the dentate line? The whitehead hemorrhoidectomy. Am J Surg. 1988;156: 182.
92. Wolff BG, Culp CE. The Whitehead hemorrhoidectomy: an unjustly maligned procedure. Dis Colon Rectum. 1988;31:587.
93. Barrios G, Khubchandani M. Whitehead operation revisited. Dis Colon Rectum. 1979;22:330.
94. Longo A. Treatment of hemorrhoidal disease by reduction of mucosal and hemorrhoidal prolapse with circular suturing device: a new procedure. In: Proceedings of the 6th world congress of Endoscopic Surgery. Bologna: Monduzzi Editore; 1998. p. 777–84.
95. Tjandra JJ, Chan MK. Systematic review on the procedure for prolapse and hemorrhoids (stapled hemorrhoidopexy). Dis Colon Rectum. 2007;50:878–92.
96. Mehigan BJ, Monson JR, Hartley JE. Stapling procedure for haemorrhoids versus Milligan-Morgan Haemorrhoidectomy: randomized controlled trial. Lancet. 2000;355:782–5.
97. Smyth EF, Baker RP, Wilken BJ, Hartley JE, White TJ, Monson JR. Stapled versus excision hemorrhoidectomy: long term follow up of a randomized controlled trial. Lancet. 2003;361:1437–8.
98. Rowsell M, Bello M, Hemingway DM. Circumferential mucosectomy (stapled haemorrhoidectomy) versus conventional haemorrhoidectomy: randomized controlled trial. Lancet. 2000;355: 779–81.
99. Au-Yong M, Rowsell M, Hemingway DM. Randomised controlled clinical trial of stapled haemorrhoidectomy vs. conventional haemorrhoidectomy; a three and a half year follow up. Colorectal Dis. 2004;6:37–8.

100. Ho YH, Cheong WK, Tsang C, et al. Stapled hemorrhoidectomy cost and effectiveness. Randomized, controlled trial including incontinence scoring, anorectal manometry, and endoanal ultrasound assessments at up to three months. Dis Colon Rectum. 2000;43:1666–75.
101. Ooi BS, Ho YH, Tang CL, Eu KW, Seow-Choen F. Results of stapling and conventional hemorrhoidectomy. Tech Coloproctol. 2002;6:59–60.
102. Helmy MA. Stapling procedure for hemorrhoids versus conventional hemorrhoidectomy. J Egypt Soc Parasitol. 2000;30:951–8.
103. Boccasanta P, Capretti PG, Venturi M, et al. Randomised controlled trial between stapled circumferential mucosectomy and conventional circular hemorrhoidectomy in advanced hemorrhoids with external mucosal prolapse. Am J Surg. 2001;182:64–8.
104. Ganio E, Altomare DF, Gabrielli F, Milito G, Canuti S. Prospective randomized multicentre trial comparing stapled with open haemorrhoidectomy. Br J Surg. 2001;88:669–74.
105. Shalaby R, Desoky A. A randomized clinical trial of stapled versus Milligan-Morgan haemorrhoidectomy. Br J Surg. 2001;88:1049–53.
106. Pavlidis T, Papaziogas B, Souparis A, Patsas A, Koutelidakis I, Papaziogas T. Modern stapled Longo procedure vs. conventional Milligan-Morgan hemorrhoidectomy: a randomized controlled trial. Int J Colorectal Dis. 2002;17:50–3.
107. Hetzer FH, Demartines N, Handschin AE, Clavien PA. Stapled vs excision hemorrhoidectomy: long-term results of a prospective randomized trial. Arch Surg. 2002;137:337–40.
108. Correa-Rovelo JM, Tellez O, Obregon L, Miranda-Gomez A, Moran S. Stapled rectal mucosectomy vs. closed hemorrhoidectomy: a randomized, clinical trial. Dis Colon Rectum. 2002;45:1367–74.
109. Wilson MS, Pope V, Doran HE, Fearn SJ, Brough WA. Objective comparison of stapled anopexy and open hemorrhoidectomy: a randomized, controlled trial. Dis Colon Rectum. 2002;45: 1437–44.
110. Ortiz H, Mazo J, Armendariz P. Randomized clinical trial of stapled hemorrhoidopexy versus conventional diathermy haemorrhoidectomy. Br J Surg. 2002;89:1376–81.
111. Kairaluoma M, Nuorva K, Kellokumpu I. Day-case stapled (circular) vs. diathermy hemorrhoidectomy: a randomized, controlled trial evaluating surgical and functional outcome. Dis Colon Rectum. 2003; 46:93–9.
112. Palimento D, Picchio M, Attanasio U, Lombardi A, Bambini C, Renda A. Stapled and open hemorrhoidectomy: randomized controlled trial of early results. World J Surg. 2003;27:203–7.
113. Picchio M, Palimento D, Attanasio U, Renda A. Stapled vs open hemorrhoidectomy: long-term outcome of a randomized controlled trial. Int J Colorectal Dis. 2006;21:668–9.
114. Cheetham MJ, Cohen CR, Kamm MA, Phillips RK. A randomized, controlled trial of diathermy hemorrhoidectomy vs. stapled hemorrhoidectomy in an intended day-care setting with longer-term follow-up. Dis Colon Rectum. 2003;46:491–7.
115. Krska Z, Kvasnieka J, Faltyn J, et al. Surgical treatment of haemorrhoids according to Longo and Milligan-Morgan: an evaluation of postoperative tissue response. Colorectal Dis. 2003;5:573–6.
116. Racalbuto A, Aliotta I, Corsaro G, Lanteri R, Di Cataldo A, Licata A. Hemorrhoidal stapler prolapsectomy vs Milligan-Morgan hemorrhoidectomy: a long-term randomized trial. Int J Colorectal Dis. 2004;19:239–44.
117. Senagore AJ, Singer M, Abcarian H, Procedure for Prolapse and Hemorrhoids (PPH) Multicenter Study Group, et al. A prospective, randomized, controlled multi-center trial comparing stapled hemorrhoidopexy and Ferguson hemorrhoidectomy: perioperative and one-year results. Dis Colon Rectum. 2004;47:1824–36.
118. Lau PY, Meng WC, Yip AW. Stapled haemorrhoidectomy in Chinese patients: a prospective randomized controlled study. Hong Kong Med J. 2004; 10:373–7.
119. Bikhchandani J, Agarwal PN, Kant R, Malik VK. Randomized controlled trial to compare the early and mid-term results of stapled versus open hemorrhoidectomy. Am J Surg. 2005;189:56–60.
120. Basdanis G, Papadopoulos VN, Michalopoulos A, Apostolidis S, Harlaftis N. Randomized clinical trial of stapled hemorrhoidectomy vs. open with Ligasure for prolapsed piles. Surg Endosc. 2005; 19:235–9.
121. Ortiz H, Marzo J, Armendariz P, De Miguel M. Stapled hemorrhoidopexy vs. diathermy excision for fourth- degree hemorrhoids: a randomized, clinical trial and review of the literature. Dis Colon Rectum. 2005;48:809–15.
122. Chung CC, Cheung HY, Chan ES, Kwok SY, Li MK. Stapled hemorrhoidopexy vs. harmonic scalpel hemorrhoidectomy: a randomized trial. Dis Colon Rectum. 2005;48:1213–9.
123. Gravie JF, Lehur PA, Huten N, et al. Stapled hemorrhoidopexy versus Milligan-Morgan hemorrhoidectomy: a prospective, randomized, multicenter trial with 2-year postoperative follow up. Ann Surg. 2005;242:29–35.
124. Kraemer M, Parulava T, Roblick M, Duschka L, Muller- Lobeck H. Prospective, randomized study: proximate PPH stapler vs. ligasure for hemorrhoidal surgery. Dis Colon Rectum. 2005;48: 1517–22.
125. Boccasanta P, Venturi M, Roviaro G. Stapled transanal rectal resection versus stapled anopexy in the cure of hemorrhoids associated with rectal prolapse. A randomized controlled trial. Int J Colorectal Dis. 2007;22:245–51.
126. Renzi A, Brillantino A, Di Sarno G, et al. PPH-01 versus PPH-03 to perform STARR for the treatment

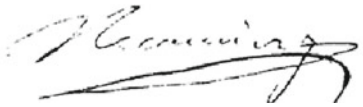
of hemorrhoids associated with large internal rectal prolapse: a prospective multicenter randomized trial. Surg Innov. 2011;18:241–7.
127. Corman ML, Carriero A, Hager T, et al. Consensus conference on the stapled transanal rectal resection (STARR) for disordered defaecation. Colorectal Dis. 2006;8:98–101.
128. Cheetham MJ, et al. Persistent pain and fecal urgency after stapled hemorrhoidectomy. Lancet. 2000;356:730–3.

Benign Disease, Fissures, Presentation and Pathophysiology

15

Mark T.C. Wong and Francis Seow-Choen

Presentation



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An anal fissure is a linear ulcer in the squamous epithelium of the anal canal located just distal to the dentate line. The French surgeon, Joseph-Claude-Anthelme Recamier (1774–1852), has been widely credited for having first described this pathology in the 1820s and subsequently went on to describe the technique of anal dilatation for treating this condition [1].

Anal fissures occur in both sexes equally, commonly affecting young and otherwise healthy individuals. However, the true incidence is likely to be higher, due to the combination of many people not seeking medical attention and with many others achieving resolution without intervention, particularly in the case of superficial anal fissures. Nonetheless, the predominant symptom of pain is often distressing enough to compel patients to seek medical attention, making anal fissures one of the most common anorectal pathologies treated by the colorectal surgeon, accounting for between 6.2 and 15 % of visits to colorectal clinics [2–6].

Sir Alan Parks once wrote that ‘if a patient complains of anal pain, the chances are that he has a fissure’ [7]. The pain is typically excruciating and has been described as the ‘passing of broken glass’, only to be followed by a burning pain that can persist for many hours afterwards. Goligher in turn described this lesion as causing ‘an amount of suffering out of proportion to the size of the lesion’ [8]. Not surprisingly, it had been shown that the pain associated with an anal fissure can result in a significant deterioration in the quality of life of affected patients [9]. There

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are often other accompanying symptoms such as the passing of bright red blood as well as anal pruritus. The fissure should be visualised by parting the buttocks and anal verge only, as this is often impeded by anal pain. The fissure can almost always be seen and the diagnosis secured. Digital or proctoscopic examination, however, is often impossible due to extreme discomfort and should not be attempted in such situation anyway. There is no immediate reason to perform an examination under general anaesthesia to confirm the diagnosis under normal circumstances especially in a young and otherwise healthy person. Table 15.1 gives the common symptoms reported in studies on anal fissures.

Classification of Fissures

Superficial vs Deep

Anal fissures are classically and usually classified into acute or chronic fissures according to well-established morphological features. The length of

Table 15.1 Common symptoms in patients with anal fissure

S/N	Study	Year	Symptoms	No of patients (%)
1.	Jensen SL	1986 (*BMJ*)	Post-defaecatory pain	96 (100)
			Bleeding	74 (77.1)
			Constipation	65 (67.7)
			Pruritus	36 (37.5)
			Soiling	17 (17.7)
			Diarrhoea	8 (8.3)
2.	Maria G	1998 (*NEJM*)	Post-defaecatory pain	30 (100)
			Nocturnal pain	4 (13.3)
3.	Maria G	1998 (*Annals*)	Post-defaecatory pain	57 (100)
			Nocturnal pain	16 (28.1)
4.	Brisinda G	1999 (*NEJM*)	Post-defaecatory pain	50 (100)
			Nocturnal pain	9 (18)
			Bleeding	9 (18)
5.	Kocher HM	2002 (*BJS*)	Post-defaecatory pain	60 (100)
			Bleeding	46 (76.7)
			Pruritus	44 (73.3)
			Discharge	25 (41.7)
6.	Arroyo A	2004 (*JACS*)	Post-defaecatory pain	74 (92.5)
			Bleeding	66 (73.3)
			Constipation	53 (66.3)
			Pruritus	43 (53.8)
7.	Arroyo A	2005 (*Am J Surg*)	Post-defaecatory pain	73 (91.3)
			Bleeding	67 (83.8)
			Constipation	55 (68.8)
			Pruritus	43 (53.8)
8.	Ho KS	2005 (*BJS*)	Post-defaecatory pain	94 (71.2)
			Bleeding	69 (52.3)
			Pruritus	33 (25)
9.	Brisinda G	2007 (*BJS*)	Post-defaecatory pain	100 (100)
			Nocturnal pain	16 (16)
			Bleeding	23 (23)
10.	Renzi A	2008 (*DCR*)	Post-defaecatory pain	49 (100)
			Bleeding	49 (100)

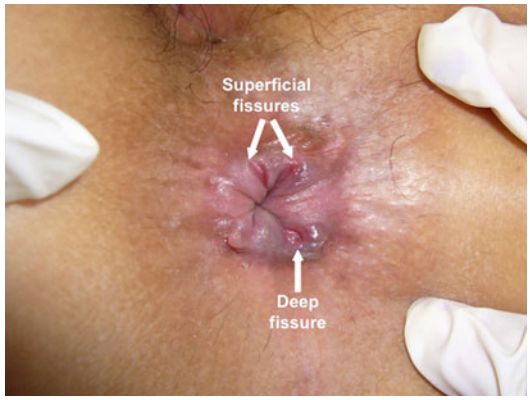

Fig. 15.1 Superficial and deep anal fissure

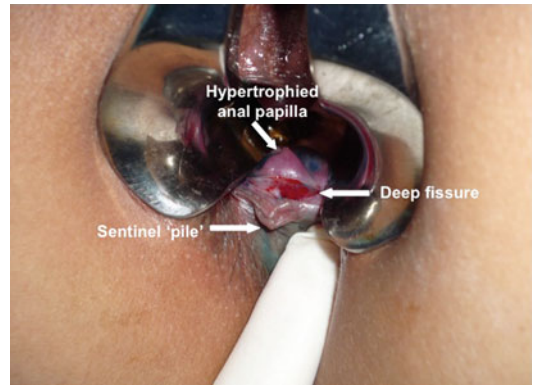

Fig. 15.2 Deep anal fissure with hypertrophied anal papilla and sentinel pile

time of symptoms ironically is usually irrelevant to whether a fissure is acute or chronic. These names are therefore inaccurate and misleading. We suggest a complete name change so as not to perpetuate this mistake. As such, we prefer to call 'acute fissures' superficial fissures and 'chronic anal fissures' deep anal fissures. Superficial fissures, as the name implies, involve only the superficial mucocutaneous layers of the anal canal. They may have symptoms of severe pain and bleeding, presenting as a superficial separation of the anoderm, usually linear or elliptical-shaped in appearance with sharply demarcated edges (Fig. 15.1). The vast majority of superficial fissures will heal spontaneously within days or at most up to 6 weeks of appropriate conservative treatment [10]. In contrast, deep fissures persist often and either tend not to heal without intervention or recur regularly.

Deep anal fissures are recognised by most authors by the morphological features of a deep and wide pear-shaped ulcer often with visible fibres of the internal anal sphincter and minimal granulation tissue at its base. Other features of a deep anal fissure include the distinctive triad of indurated ulcer edges, a distal skin tag (sentinel pile) and a proximal hypertrophied anal papilla [4, 11] (Fig. 15.2). The latter however is not universal. A standardised definition should be established to facilitate the uniformity of future trials and to explain the widely differing healing rates of the various treatments in the literature. Lindsey et al. [11] have proposed a definition based on the combination of chronology and morphology by describing a deep anal fissure as 'the presence of visible transverse internal anal sphincter fibres at the base of an anal fissure of duration not less than 6 weeks'. However, we have often observed such features presenting within a week or less of painful defaecation and/or bleeding, and therefore, we stress that duration is irrelevant and again urge a more appropriate name for these sorts of fissures to be deep anal fissures. In addition, deep fissures may be further defined by the presence or absence of the hypertrophied anal papilla. Our own observations lead us to postulate that a superficial fissure is a shallow separation of the anoderm that does not reach the internal sphincter muscle. This sort of fissure heals readily without too much fuss. Most of us would have one time or the other have had this sort of anal pain but the majority of these heal spontaneously. Deep anal fissures however may result from prolongation or repeat of the forces causing superficial anal fissures, or else, they may occur de novo as a result of a deep tearing right from the start and may therefore appear almost immediately. An example of such a fissure would be a deep fissure appearing at the site of a haemorrhoidectomy wound.

Typical vs Atypical

Fissures can also be classified based on their location and aetiology. Typical fissures are

usually single and situated in the posterior midline (6 o'clock position), with 2.5–13 % of fissures occurring in the anterior midline (12 o'clock position) in up to 10 % of women compared to 1 % of men [4, 8, 12]. Occasionally, patients can also present with fissures in both these locations concurrently. Multiple fissures occurring in unusual locations should raise suspicion of an atypical aetiology.

Atypical or secondary fissures are usually associated with some underlying diseases, most notably inflammatory bowel diseases, human immunodeficiency virus (HIV) infection, anal cancer, syphilis, sarcoidosis, tuberculosis and radiotherapy, or may even point to possible sexual abuse and anal-receptive intercourse. Such fissures tend to be accompanied by an unusual history or clinical features such as hypotonic anal sphincters, large size, irregularity as well as being multiple or sited at unusual lateral locations or both [4, 13, 14]. It is therefore essential to exclude more proximal bowel or systemic pathology in the presence of atypical features on clinical examination, which generally requires an examination under anaesthesia and even biopsy to establish a conclusive diagnosis. In addition, the persistence of fissures following surgical sphincterotomy should also raise one's suspicion of a possible secondary aetiology.

The incidence of deep anal fissures in patients with Crohn's disease has been reported to be approximately 26 % and is sometimes the only manifestation of the disease [15, 16]. They tend to occur in atypical locations, are often deeper and can potentially result in significant deformity. In addition, they are commonly associated with other anorectal pathology, especially fistula. Lewis et al. reported that 5 of 21 patients with persistent fissures after surgical sphincterotomy were found to have Crohn's disease and the occurrence of fissures has been postulated to be due to microvascular ischaemia.

The anorectum is one of the sites commonly afflicted by HIV infection, which may manifest as several lesions including anal fissures [17–21]. The incidence of anal fissures in HIV patients is reported to vary between 7 and 32 %, with about 5 % of patients presenting without foreknowledge of the diagnosis of HIV infection [21, 22]. Anal fissures in such patients are usually located more proximally, often extending above the dentate line and located at atypical lateral sites [23–25]. These fissures are often associated with other anorectal lesions, such as ulcerations, perianal abscesses and fistulae, with concurrent infections variously attributed to Cryptococcus sp., cytomegalovirus (CMV), Chlamydia sp., herpes simplex virus (HSV), Treponema pallidum and Haemophilus ducreyi [20, 26–30]. It should be noted that anal fissures in HIV patients may be associated with hypotonicity of the anal sphincter, as a result of persistent diarrhoea and their anal-receptive sexual practices [21, 25]. As such, surgical intervention is not routinely recommended as they are commonly associated with impaired anal sphincter function.

Anorectal tuberculosis is exceedingly rare, occurring in approximately 0.7 % of patients infected with this granulomatous disease. Adding to the diagnostic challenge is the notoriously low yield of acid-fast bacilli (AFB) from the lesion [23, 29, 30]. The ulcerated form of anal TB typically presents as a superficial, non-indurated lesion, with a hemorrhagic necrotic base that is granular and covered with thick mucopurulent secretions. The lesion may be very painful or the patient may have few symptoms [31]. The tuberculin skin test remains a valuable guide because it is positive in 75 % of cases. Patients with a tuberculous fissure will usually have symptoms of the disease in other systems, especially in the chest and other parts of the gastrointestinal tract, either as an extension of the original lesion or due to its spread via the lymphatics [32]. In addition, tuberculosis can be a complicating infection of HIV-positive patients, with the incidence and severity of ano-perianal tuberculosis increasing due to the increasing incidence of HIV infection [33, 34]. The diagnosis may be suspected from the persistence of anal fissure despite conventional therapy, and confirmation is by tissue biopsy.

Other causes of secondary anal fissures include malignant lesions such as various forms of leukaemia, lymphoma as well as squamous and basal cell carcinomas, malignant melanoma, Kaposi's sarcoma and adenocarcinoma of the

anorectum, all of which may be diagnosed based on histology obtained from biopsies of the lesion [25, 30].

High Pressure vs Low Pressure

Since the seminal lecture by Brodie BC in 1835 regarding the existence of elevated anal tone in patients with anal fissures, anal hypertonicity had remained a leading hypothesis and had helped to shape much of the research and development of treatment for this disease entity [35]. Table 15.2 shows the distribution of anal canal resting pressures in studies of patients with anal fissures before and after treatment.

Nevertheless, debate still ensues regarding the consistency of this association, as anal fissures associated with normal or hypotonic sphincters (low-pressure fissures) have been reported in the postpartum period, in the elderly as well as in secondary fissures associated with underlying disease [14, 36, 37]. Bove and colleagues, in a study assessing anal pressure profiles of patients with deep anal fissure, revealed that 52.1 % of patients had mean resting anal pressures within the normal range (35–50 mmHg), with up to 5 % of patients (significantly older of mean age 56.5 ± 11.1 years) having a hypotonic anal canal [38]. These findings highlight the fact that sphincter hypertonicity is not universally present in patients with anal fissures and that treatment should not just be directed at decreasing anal canal pressures.

Considered the gold standard against which all treatments are compared, lateral internal

Table 15.2 Anal canal pressures in patients with deep anal fissures

Study	Year	No. of patients	Treatment	Mean anal resting pressure ± SD (mmHg)	
				Before	After
Olsen J	1987 (*IJCD*)	10	Lateral anal sphincterotomy	80 ± 10	68 ± 5
		10	vs anal dilatation	84 ± 4	55 ± 13[a]
Lund JN	1997 (*Lancet*)	38	0.2 % GTN ointment	85.2 ± 23.2	55.8 ± 22.1[a]
		39	vs placebo	86.8 ± 32.8	82 ± 23.5
Maria G	1998 (*NEJM*)	15	20U botulinum toxin	109 ± 8	81 ± 8[a]
		15	vs 0.4 ml saline	102 ± 6	97 ± 7
Maria G	1998 (*Annals*)	23	15U botulinum toxin	94 ± 35	79 ± 33
		34	vs 20U botulinum toxin	110 ± 30	79 ± 27[a]
Brisinda G	1999 (*NEJM*)	25	GTN ointment	83.4 ± 15.0	71.9 ± 17[a]
		25	vs 20U botulinum toxin	89.8 ± 21.2	64.2 ± 14.9[a]
Arroyo A	2004 (*JACS*)	40	Open lateral sphincterotomy	109 ± 29	75 ± 17[a]
		40	vs closed lateral sphincterotomy	119 ± 21	78 ± 20[a]
Parellada C	2004 (*DCR*)	27	0.2 % GTN ointment	58.8 ± 14.3	41.2 ± 18[a]
		27	vs lateral sphincterotomy	52.9 ± 10.2	35.3 ± 7.4[a]
Arroyo A	2005 (*Am J Surg*)	40	Open lateral sphincterotomy	109 ± 29	72 ± 17[a]
		40	vs 25U botulinum toxin	114 ± 26	92 ± 26[a]
Ho KS	2005 (*BJS*)	48	Lateral sphincterotomy	53.1 ± 4.0	43.4 ± 4.0[a]
		43	vs tailored sphincterotomy	66.7 ± 7.6	47.7 ± 3.4[a]
		41	vs oral nifedipine	44.9 ± 4.5	42.7 ± 6.8
Brisinda G	2007 (*BJS*)	50	20U botulinum toxin	90.2 ± 19.7	70.2 ± 18.9[a]
		50	vs 0.2 % GTN ointment	89.2 ± 21.0	72.6 ± 16.0[a]
Renzi A	2008 (*DCR*)	24	Pneumatic dilatation	94.4 ± 11.3	65.6 ± 6.6[a]
		25	vs lateral sphincterotomy	96 ± 12.1	63 ± 5.7[a]

Normal resting anal pressure, 50–70 mmHg; maximum voluntary (squeeze) pressures were normal in all studies with no significant change after treatment; *GTN* glyceryl trinitrate, *ISDN* isosorbide dinitrate

[a]Significant changes in pressure compared to baseline values (i.e. $p < 0.05$)

sphincterotomy has been shown to effectively decrease resting anal pressure by up to 50 %, helping to improve anodermal blood flow and thereby possibly promoting fissure healing in patients with elevated sphincter pressures [39, 40]. However, lateral anal sphincterotomy had been shown to be associated with anal incontinence postoperatively, possibly as a result of overzealous sphincter division or due to the presence of undiagnosed sphincter injuries, with incontinence rates ranging from 3.3 to 16 % [41–48]. Several authors have shown encouraging results in their efforts to reduce incontinence rates by using a tailored approach to sphincterotomy guided by preoperative manometric findings [49, 50] as well as by limiting the extent of division to the fissure apex [51]. Consequently, it is easy to appreciate the disastrous consequences of an unnecessary sphincterotomy performed on a patient with normal or low sphincter pressures. In the latter groups of patients, a sphincter-preserving approach using anal advancement flaps offers a safer alternative, with several studies reporting favourable healing rates of more than 80 %, without the morbidity of postoperative incontinence [52–55].

Pathophysiology

Despite having been described more than 180 years ago, the precise mechanisms surrounding the pathophysiology of anal fissures had yet to be fully unravelled. However, with improvement in the understanding of the physiology of the anal sphincter, significant progress has been made in recent years and this has provided the rationale for current treatment modalities. An understanding of anal sphincter physiology is therefore an essential prelude to any discussion regarding the current treatment options for anal fissures.

Internal Anal Sphincter Physiology

The human internal anal sphincter (IAS) exists in a state of tonic contraction and its function is modulated by three main components, namely, intrinsic myogenic tone and the enteric and autonomic nervous systems, reflecting the sphincter specialisation of this muscle [56]. Basal tone of the IAS is primarily myogenic, which is modulated by neurohormonal substances such as alpha- and beta-adrenoreceptor agonists, as well as inhibitory neurotransmitters including nitric oxide (NO), vasoactive intestinal peptide (VIP) and carbon monoxide (CO) [57, 58].

Firstly, intrinsic myogenic tone is a spontaneous phenomenon that results from the contraction of smooth muscle cells within the IAS, mediated by the influx of calcium through L-type calcium channels [59]. There are also two groups of alpha (α) adrenoreceptors within the IAS; stimulation of α1-adrenoreceptors causes IAS contraction [60], whilst activation of α2-adrenoreceptors inhibits non-adrenergic, non-cholinergic (NANC) relaxation. Conversely, the stimulation of beta-adrenoreceptors inhibits sympathetic stimulation of the IAS [57]. Consequently, the dominant α-adrenoreceptor population in the IAS results in the tonic state of contraction.

The enteric nervous system is located in the Auerbach's and Meissner's plexuses in the wall of the gut and is responsible for peristalsis as well as local reflexes such as the rectoanal inhibitory reflex. These nerves are known to be NANC because neither guanethidine nor atropine blocks their activity, although the neurotoxin tetrodotoxin blocks their activity [61]. Activation of α2-adrenoreceptors in the myenteric inhibitory neurons presynaptically inhibits NANC relaxation. Relaxation is mediated through directly decreasing intracellular calcium concentration as well as increasing cyclic guanosine monophosphate and cyclic adenosine monophosphate. Potassium influx hyperpolarises the cell membrane and decreases calcium entry. In addition, inhibitory neurotransmitters, such as nitric oxide and vasoactive intestinal peptide, mediate NANC relaxation.

Nitric oxide is the major neurotransmitter mediating NANC relaxation of the IAS [57, 58], an action that is blocked by *N*-nitro-L-arginine (a nitric oxide synthase inhibitor) and enhanced by L-arginine (a nitric oxide precursor) [62]. The

presence of nitric oxide synthase (NOS)-positive neurons in the rectal myenteric plexus and anal canal has been confirmed by immunohistochemical studies, demonstrating that these neurons ramify throughout the IAS and lay in close proximity to smooth muscle cells [63]. In Hirschsprung's disease, a condition in which the rectoanal inhibitory reflex is absent, nerves containing NOS are absent from the non-relaxing segment but present in the normal segment of gut [64].

The third influence is the autonomic nervous system, which affects contraction and relaxation of the internal sphincter via sympathetic and parasympathetic postganglionic fibres, respectively. The IAS receives sympathetic innervation from the fifth lumbar segment via the hypogastric nerves. It is not entirely clear precisely how these fibres operate, but they may act directly on the smooth muscle, indirectly on the nerves of the enteric nervous system or both [11]. There is a relative dominance of sympathetic over parasympathetic neural input, leading to a background tonic state of contraction of the internal sphincter.

A Historical Perspective of the Aetiology of Anal Fissure

An early theory of Ball suggested that the passage of a hard faecal bolus tore down an anal valve, leaving oedematous coiled-up skin at the anal verge in the form of a sentinel pile [65]. However, the principle shortcoming of this theory is that an intact anal valve is usually present above a fissure, with the possibility of even a hypertrophied anal papilla being present in deep fissures. Furthermore, the latter theory fails to explain the preponderance of fissures in the posterior midline of the anal canal, as well as the fact that fissures rarely extend up beyond the dentate line as they would if an anal valve had been torn down.

Miles suggested that varicose veins of the anal canal were implicated in both the formation and persistence of anal fissures, which resulted in a 'pecten band' in the distal anal canal [66]. This band was believed to render the overlying anal mucosa more prone to tearing and was conversely suggested that anal fissures rarely developed in the absence of such a band [67]. Thus, division of this 'pecten band' was advised as treatment. Eisenhammer however showed that this band was in fact the hypertonic IAS and that division of this structure amounted to a sphincterotomy [68]. Building upon the theory of regional varicosities, Rankin and colleagues proposed that the fissure was a form of varicose ulcer [69]. They suggested that a fissure was initiated by trauma from a faecal mass or sepsis in the anal crypts and was made intractable by phlebitis in the underlying varicosities. The posterior location of most fissures was explained by the susceptibility of the anorectal angle to trauma during defaecation. However, this concept fell out of favour due to the fact that fissures rarely reach the level of the dentate line, let alone the anorectal angle.

In trying to explain the preponderance of fissures in the posterior midline, Blaisdell drew reference to the anatomy of the external anal sphincter. He explained that the bulk of the external sphincter muscle did not completely encircle the anal canal but instead decussated to form a Y shape posteriorly, with only the distal subcutaneous portion forming a complete ring. He postulated that at the junction of these two parts of the external sphincter, the overlying anal mucosa was poorly supported and more prone to tearing from the passage of a large faecal bolus [70]. Blaisdell thus advocated the division of the subcutaneous part of the external sphincter, which became known as the 'Blaisdell bar', with subsequent works by Milligan and Gabriel supporting this concept [71, 72].

The eventual work of Eisenhammer drew attention to the role of the internal anal sphincter, in which he described the presence of spasms of this muscle in patients with fissures, associated with contracture of the anal canal and rigidity of the overlying mucosa [68]. It was then thought that the exposed muscle fibres often seen at the base of deep fissures were in fact part of the internal sphincter and not the external sphincter as originally thought. Brown and colleagues went on to show that the IAS of patients with anal

fissures had comparatively more fibrosis on histological assessment, compared with patients undergoing anorectal excision for rectal cancer who had otherwise normal anal canals [73]. It was therefore postulated that a myositis occurs early in the course of a fissure and that this is the underlying cause of both spasm and fibrosis. However, the amount of fibrosis did not correlate with the duration of symptoms and the cause of the fibrosis remains uncertain.

A Unifying Concept for the Aetiology of Deep Anal Fissure

The present understanding of anal fissures is fragmented and hazy. Many authors, in elucidating and attempting to understand anal fissure, have failed to differentiate the factors that initiate acute anal tears against factors that perpetuate anal fissures or cause them to extend and deepen. We believe that in order to really understand the problem of anal fissures, we need to look separately at initiating and perpetuating causes of anal fissures.

Initiating Factors

Central to the understanding of fissure formation is the concept of perineal descent, which can result in overstretching of the overlying anal mucosa, making it prone to subsequent trauma by initiating factors like a hard faecal bolus, explosive diarrhoea or a foetus.

Perineal Descent as an Initiating Cause of Anal Fissure

Based on the 'levator complex' theory proposed by Shafik, it was initially believed that the levator ani contracted and shortened during defaecation, resulting in its elevation and lateral retraction and consequent opening of the anal canal [74–76]. However, Guo et al. [77] have subsequently shown that during defaecation, the increase in intra-abdominal pressure in fact pushes down the levator ani without associated shortening of the levator ani muscles. This lengthening and descent of the levator ani stretches the muscle and may be the reason for the tearing that occurs at the anal verge [77]. Dynamic computed tomographic (CT) defaecography has further confirmed that during defaecation in normal individuals, the levator ani descends by as much as 20 mm, changing from a funnel shape at rest to a deeper basin shape; the resultant force on the levator ani pushes the muscle downwards which then widens the genital hiatus and opens the anus for the passage of stool [77] (see Fig. 15.3). In certain individuals, particularly multiparous or elderly

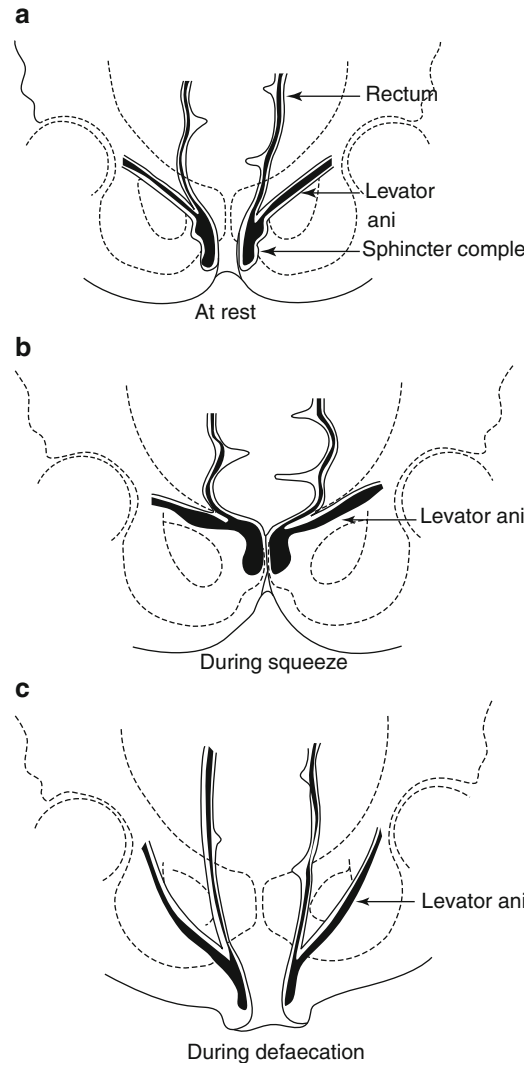

Fig. 15.3 Changes in the levator ani during defaecation. (**a**) At rest, levator ani is 'funnel'-shaped. (**b**) During squeeze, levator ani ascends and becomes 'plate'-shaped, and anus closes. (**c**) During defaecation, levator ani descends and becomes 'basin'-shaped, and anus opens

women, the perineum and levator plate can descend even more excessively during the process of defaecation as a result of the trauma of childbirth on the pelvic musculature with associated pudendal neuropathy, exacerbated by the effects of oestrogen withdrawal during menopause which can further weaken the pelvic musculature.

We believe this downward lengthening of the levator ani allows the excessive stretching of the overlying anoderm, making it more vulnerable to tearing by a hard faecal bolus or explosive diarrhoea. In particular, this overstretching tends to occur in the posterior midline and especially at the overlying anoderm due to the hard faecal bolus shearing the loose anoderm over the relatively fixed anococcygeal ligaments [78–80].

Therein lies the premise behind the use of manual perineal support during defaecation. This concept, whilst new in the understanding of anal fissures, has actually been used by many patients with obstructed defaecation. These patients instinctively use this manoeuvre to facilitate the passage of stool by pressing on the posterior perineum during defaecation [81]. Even in infants who suffer from hard bulky stools that are impacted at the anal verge, gentle pressure at the posterior perineal region may be a useful method to help evacuate the stool. However, posterior perineal support during defaecation is not easily obtained whilst sitting on a toilet seat. This has led to our own research into the reduction of trauma during defaecation, with a recent study looking into the use of a novel posterior perineal support device incorporated into a toilet seat, as a non-invasive method of improving the healing rates of deep anal fissures [80].

The premise for this device is to counteract the downward stretch of the anoderm as a result of puborectalis lengthening during defaecation that leads to shearing of the anoderm by the exiting faecal bolus. The posterior perineal support holds up the anococcygeal region just behind the posterior aspect of the anus, providing counter support at the posterior aspect of the pelvic floor behind the anus, thereby balancing the pressure exerted by faeces on the anal wall and thus reducing the stretching tension on the pelvic floor and

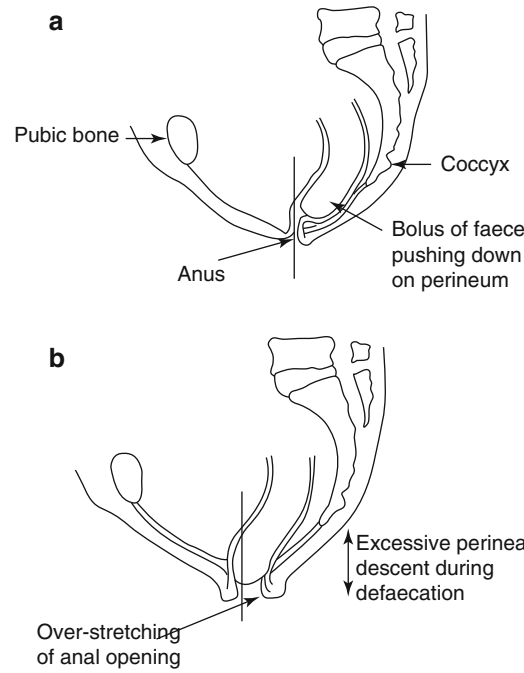

Fig. 15.4 (**a**, **b**) Faeces causing overstretching of anal opening (unsupported pelvic floor)

posterior perineal tissue (see Figs. 15.4 and 15.5). This is believed to also enhance the stimulus bringing about the reflex of defaecation, leading to more effective voiding of stool and reducing the need for straining. This preliminary study on 32 patients with symptomatic deep anal fissures revealed a 97.5 % improvement in pain, 65.6 % improvement in bleeding, 84.4 % improvement in constipation symptoms and 68.8 % improvement in abdominal discomfort after 3 months of usage. Anecdotally, we have also observed that the device has also helped to reduce pain and enhance the recovery of episiotomy wounds in the postpartum period, likely by preventing excessive perineal descent and thus limiting the stretching of the wound.

Constipation and Diarrhoea

Historically, constipation and the passage of hard stools were believed to be the main causative factor of anal fissure. Contrary to this belief, it has subsequently been shown that a history of constipation is elicited in only 13.8–25 % of patients, with another 4–7 % of patients shown to have

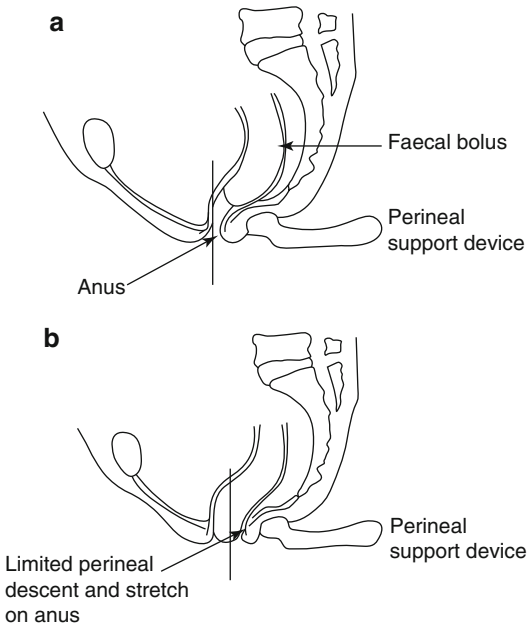

Fig. 15.5 (**a**, **b**) Perineal support prevents overstretching of anal opening (supported pelvic floor)

diarrhoea as a predisposing factor instead [82–84]. As such, we are convinced that even diarrhoea, when sufficiently explosive or persistent, is able to result in tearing of the distal anoderm, setting up the vicious cycle of anal spasms and further trauma. In addition, patients with fissures on the background of chronic or persistent diarrhoea should raise the possibility of an underlying secondary pathology such as HIV infection, Crohn's disease or tuberculosis and all attempts must be made to exclude the above aetiologies so as to guide effective treatment.

Fissures in Pregnancy

Together with haemorrhoidal disease, anal fissures affect approximately one-third of females in the postpartum period, with concurrent constipation being an important risk factor [85]. Anal fissures may be associated with childbirth in between 3 and 11 % of patients and commonly occur in the anterior midline, with the risk increasing in traumatic or instrumental deliveries [82, 86, 87]. The resulting incidence of fissures thus increases from approximately 1.2 % during pregnancy to 15.2 % in the early postpartum period [85]. The combination of the forces associated with the strong bearing down during delivery and the shearing force from the foetal head on the anal mucosa and the relative laxity of structural tethering after childbirth render the anoderm more susceptible to tearing trauma. In women, the anal canal is hence more vulnerable to being pushed excessively by the passage of hard stools, especially when the perineal body has been previously traumatised by childbirth. This could also explain why women have more frequent anterior fissures and rectocele compared with men. In a study by Corby and colleagues, the authors found that 9 % of primigravid women developed postpartum anal fissure; anal manometry further revealed a decrease in anal canal resting and squeeze pressures in the postpartum period, reiterating the importance of nonsurgical management or sphincter-preserving procedures in these situations in order to avoid debilitating incontinence, the risk of which is increased by the presence of concurrent obstetric sphincter injuries [36].

The initiating factor therefore is sufficient and repeated trauma during the act of defaecation leading to a deep tear in the anal canal associated with a relative lack of superficial anodermal support especially posteriorly where the anococcygeal ligaments limit the flexibility of the anal sphincters. This sort of tear is associated with straining to pass motion and diarrhoea and may be associated with an excessively hypertonic or even hypotonic anal sphincter.

Perpetuating Factors

Once the initial tear has occurred, intrinsic anal sphincter hypertonia and relative tissue ischaemia then drive a vicious cycle of non-healing and repeated trauma that can lead to deep fissures.

Internal Anal Sphincter Hypertonia

Increased understanding of internal anal sphincter (IAS) physiology has revealed the important role anal tone plays in the pathophysiology of anal fissures. Reduced levels of nitric oxide synthase (NOS), the primary enzyme involved in nitric oxide (NO) synthesis, were demonstrated

in the IAS of patients with anal fissures compared to controls [86–88], with mean resting anal pressures noted to be often higher than 90 mmHg in patients with fissures [2, 13, 39, 40]. Furthermore, it has been shown that maximum voluntary contraction pressures of the external sphincter remain largely unchanged in both patients with anal fissures and healthy controls, suggesting that the IAS alone appears to be responsible for the hypertonia [89]. This reduced production of NO provides a possible explanation for the high anal pressures seen in most affected patients and possibly explains why pressures return to pretreatment values in patients whose fissures have healed by nonsurgical methods [90].

In addition, the hypertonia is unlikely to be secondary to pain alone, as pain relief with topical application of local anaesthetics does not alleviate the elevated pressures [91]. Similar findings of reduced NOS activity have also been reported in the gastro-oesophageal junctions of achalasia patients [92]. Furthermore, a long high-pressure zone in the anal canal with ultraslow waves is commonly seen in these patients [91]. To date, there have been no studies on the role of VIP in the aetiology of anal fissures. However, a debate still ensures as to whether the elevated pressure is a direct cause or an effect of the disease [89]. It should also be noted that a minority of anal fissures are not associated with sphincter hypertonia and these 'low-pressure' fissures will be discussed in the later sections.

Ischaemia

Over the past two decades, local ischaemia had been gaining credence as a significant perpetuating factor in deep fissures. The blood supply of the distal anal canal is via the inferior rectal arteries, branches of the internal pudendal artery. These vessels cross the ischio-rectal fossae and divisions pass through the anal sphincters to reach the mucosa. In a cadaveric study by Klosterhalfen et al. [93], post-mortem angiography of the inferior rectal artery performed in 41 subjects revealed that 85 % had a paucity of branches of the inferior rectal artery at the posterior commissure. Morphometric studies of the capillaries further revealed them to be less dense in both the sub-anodermal space and within the IAS in the posterior midline in the majority of subjects. It was thus suggested that ischaemia of the posterior midline may explain the predilection of anal fissures at this position and the lack of granulation tissue seen at the base of a deep anal fissure [93].

Schouten et al. went on to propose the concept of anal fissures being ischaemic ulcers [94]; patients with deep anal fissures and healthy volunteers underwent anal manometry and Doppler flowmetry studies, with those suffering from fissures noted to have both a significantly higher resting anal sphincter tone and decreased anodermal blood flow, compared to healthy volunteers [91]. It was further demonstrated that patients whose fissures had healed following lateral sphincterotomy conversely had a decreased anal sphincter tone and an increased blood flow to the posterior midline of the anal canal.

The above two postulates of IAS hypertonicity and relative ischaemia thus appear intertwined, with the high anal tone potentially aggravating the already poor blood supply of the posterior anal canal. This has led to the current understanding that following an initiating trauma to the anal canal in susceptible individuals with internal sphincter hypertonia, there is little or no healing of the anal mucosa due to tissue ischaemia caused by compression of the inferior rectal arteries by the internal sphincter. The paucity of blood flow prevents healing of the anal fissure until the cycle of internal sphincter hypertonia and decreased blood flow is broken by muscle relaxants or surgery.

Repeated Perineal Descend and Trauma to the Anoderm

The stretching of the puborectalis and therefore the anoderm is not just associated with the initiation of superficial anal fissures, but is present each time the afflicted patient defaecates. This repeated action on the overlying anoderm is therefore playing an important part as well in the perpetuating of the fissure causing fissures to deepen and not to heal.

The perpetuating causes leading to the formation of deep anal fissures are therefore the

hypertonicity of the anal canal due to pain as well as the repeated stretching of the anoderm due to perineal descent and the passage of stool with both this repeated trauma and ischaemia of the fissure being important factors leading to non-healing deep fissures.

References

1. Récamier JC. Extension, massage et percussion cadancee dans le traitment des contractures musculaires. Revue Médicine Francais et Estranger. 1838;1:74.
2. McCallion K, Gardiner KR. Progress in the understanding treatment of chronic anal fissure. Postgrad Med J. 2001;77:753–8.
3. Prescatori M, Interisano A. Annual report of the Italian coloproctology unit. Tech Coloproctol. 1995;3: 29–30.
4. Lund JN, Scholefield JH. Aetiology and treatment of anal fissure. Br J Surg. 1996;83:1335–44.
5. Lubowski DZ. Anal fissures. Aust Fam Physician. 2000;29:839–44.
6. Garner JP, McFall M, Edwards DP. The medical and surgical management of chronic anal fissure. J R Army Med Corps. 2002;148:230–5.
7. Parks AG. The management of fissure-in-ano. Br J Hosp Med. 1967;1:737–9.
8. Goligher JC. Surgery of the anus, rectum and colon. 5th ed. London: Bailliere Tindall; 1984. p. 150–66.
9. Griffin N, Acheson AG, Tung P, Sheard C, Glazebrook C, Scholefield JH. Quality of life in patients with chronic anal fissure. Colorectal Dis. 2004;6:39–44.
10. Jensen SL. Treatment of first episodes of acute anal fissure: a prospective randomized study of lignocaine ointment versus hydrocortisone ointment or warm sitz baths plus bran. Br Med J (Clin Res Ed). 1986;292:1167–70.
11. Lindsey I, Jones OM, Cunningham C, Mortensen NJ. Chronic anal fissure. Br J Surg. 2004;91:270–9.
12. Hananel N, Gordon PH. Re-examination of clinical manifestations and response to therapy of fissure-in-ano. Dis Colon Rectum. 1997;40:229–33.
13. Utzig MJ, Kroesen AJ, Buhr HJ. Concepts in pathogenesis and treatment of chronic anal fissure – a review of the literature. Am J Gastroenterol. 2003;98: 968–74.
14. Ayantunde AA, Debrah SA. Current concepts in anal fissures. World J Surg. 2006;30:2246–60.
15. Sweeney JL, Ritchie JK, Nicholls RJ. Anal fissure in Crohn's disease. Br J Surg. 1998;75:56–7.
16. Platell C, Mackay J, Collopy B, et al. Anal pathology in patients with Crohn's disease. Aust N Z J Surg. 1996;66:5–9.
17. Miles AJ, Mellor CH, Gazzard B, Allen-Mersh TG, Wastell C. Surgical management of anorectal disease in HIV-positive homosexuals. Br J Surg. 1990;77:869–71.
18. Scholefield JH, Northover JMA, Carr MD. Male homosexually HIV infection and colorectal surgery. Br J Surg. 1990;77:493–6.
19. Wexner SD. Sexually transmitted diseases of the colon and anus: the challenge of the nineties. Dis Colon Rectum. 1990;33:1048–62.
20. Wexner SD, Smithy WB, Milsom JW, Dailey TH. The surgical management of anorectal diseases in AIDS and pre-AIDS patients. Dis Colon Rectum. 1986;29: 719–23.
21. Nadal SR, Manzione CR, Galvão VM, Salim VR, Speranzini MB. Perianal diseases in HIV positive patients compared with a seronegative population. Dis Colon Rectum. 1999;42:649–54.
22. Barrett WL, Callahan TD, Orkin BA. Perianal manifestations of human immunodeficiency virus infection: experience in 260 patients. Dis Colon Rectum. 1998;41:606–12.
23. Brown SR, Taylor A, Adam JJ, et al. The management of persistent and recurrent chronic anal fissures. Dis Colon Rectum. 2002;4:226–32.
24. Safari A, Gottesman L, Dailey TH. Anorectal surgery in the HIV-positive patients: update. Dis Colon Rectum. 1991;34:299–304.
25. Miles AJG, Connolly GM, Barton SE, et al. Persistent ulceration of the anal margin in homosexuals with HIV infection. J R Soc Med. 1991;84:87–8.
26. Goodell SE, Quinn TC, Mkrtichian E, Schuffler MD, Holmes KK, Corey L. Herpes simplex virus proctitis in homosexual men. Clinical, sigmoidoscopic, and histopathological features. N Engl J Med. 1983;308: 868–71.
27. Consten EC, Slors JF, Danner SA, Offerhaus GJ, Bartelsman JF, Van Lanschot JJ. Local excision and mucosal advancement for anal ulceration in patients infected with human immunodeficiency virus. Br J Surg. 1995;82:891–4.
28. Candela F, Serrano P, Arriero JM, Teruel A, Reyes D, Calpena R. Perianal disease of tuberculous origin: report of a case and review of literature. Dis Colon Rectum. 1999;42:110–2.
29. Whalen Jr TV, Kovalcik PJ, Old Jr WL. Tuberculous anal ulcer. Dis Colon Rectum. 1980;23:545.
30. Smith DL, Cataldo PA. Perianal lymphoma in a heterosexual and non-immunocompromised patient: report of a case and review of literature. Dis Colon Rectum. 1999;42:952–4.
31. Le Bourgeois PC, Poynard T, Modai J, Marche C, Avril MF, Chaput JC. Peri-anal ulceration. Tuberculosis should not be overlooked. Presse Med. 1984;13:2507–9.
32. Iwase A, Shiota S, Nakaya Y, et al. An autopsy case of severe tuberculosis associated with anal fistula and intestinal perforation. Kekkaku. 1997;72:515–8.
33. Musch E, Tunnerhoff-Mucke A. Tuberculous anal fistula in acquired immunologic deficiency syndrome. Z Gastroenterol. 1995;33:440–4.
34. Kraemer M, Gill SS, Seow-Choen F. Tuberculous anal sepsis: report of clinical features in 20 cases. Dis Colon Rectum. 2000;43:1589–91.

35. Brodie BC. Lectures on diseases of the rectum; III; preternatural contraction of the sphincter ani. London Med Gaz. 1835;16:26–31.
36. Corby H, Donnelly VS, O'Herlihy C, O'Connell PR. Anal canal pressures are low in women with postpartum anal fissure. Br J Surg. 1997;84:86–8.
37. Favetta U, Amato A, Interisano A, Pescatori M. Clinical, manometric and sonographic assessment of the anal sphincters. A comparative prospective study. Int J Colorectal Dis. 1996;11:163–6.
38. Bove A, Balzano A, Perrotti P, Antropoli C, Lombardi G, Pucciani F. Different anal pressure profiles in patients with anal fissure. Tech Coloproctol. 2004;8:151–7.
39. Farouk R, Duthie GS, MacGregor AB, Bartolo DC. Sustained internal sphincter hypertonia in patients with chronic anal fissure. Dis Colon Rectum. 1994;37:424–9.
40. Schouten WR, Briel JW, Auwerda JJA, De Graaf EJ. Ischaemic nature of anal fissure. Br J Surg. 1996;83: 63–5.
41. Menteş BB, Irkörücü O, Akin M, Leventoğlu S, Tatlicioğlu E. Comparison of botulinum toxin injection and lateral internal sphincterotomy for the treatment of chronic anal fissure. Dis Colon Rectum. 2003;46:232–7.
42. Garcea G, Sutton C, Mansoori S, Lloyd T, Thomas M. Results following conservative lateral sphincterotomy for the treatment of chronic anal fissures. Colorectal Dis. 2003;5:311–4.
43. Tocchi A, Mazzoni G, Miccini M, Cassini D, Bettelli E, Brozzetti S. Total lateral sphincterotomy for anal fissure. Int J Colorectal Dis. 2004;19:245–9.
44. Liratzopoulos N, Efremidou EI, Papageorgiou MS, Kouklakis G, Moschos J, Manolas KJ, Minopoulos GJ. Lateral subcutaneous internal sphincterotomy in the treatment of chronic anal fissure: our experience. J Gastrointestin Liver Dis. 2006;15:143–7.
45. Wiley M, Day P, Rieger N, Stephens J, Moore J. Open vs closed lateral internal sphincterotomy for idiopathic fissure-in-ano: a prospective, randomized, controlled trial. Dis Colon Rectum. 2004;47:847–52.
46. Jensen SL, Lund F, Nielsen OV, Tange G. Lateral subcutaneous sphincterotomy versus anal dilatation in the treatment of fissure in ano in outpatients: a prospective randomized study. Br Med J (Clin Res Ed). 1984;289:528–30.
47. Richard CS, Gregoire R, Plewes EA, et al. Internal sphincterotomy is superior to topical nitroglycerin in the treatment of chronic anal fissure: results of a randomized, controlled trial by the Canadian Colorectal Surgical Trials Group. Dis Colon Rectum. 2000;43:1048–57.
48. Brown CJ, Dubreuil D, Santoro L, Liu M, O'Connor BI, McLeod RS. Lateral internal sphincterotomy is superior to topical nitroglycerin for healing chronic anal fissure and does not compromise long-term fecal continence: six-year follow-up of a multicenter, randomized, controlled trial. Dis Colon Rectum. 2007;50:442–8.
49. Pescatori M, Maria G, Anastasio G. "Spasm related" internal sphincterotomy in the treatment of anal fissure. A randomized prospective study. Coloproctology. 1991;1:20–2.
50. Hussein AM, Shehata MAS, Zaki Y, El Seweify MEA. Computerized anal vector manometric analysis in patients treated by tailored lateral sphincterotomy for anal fissure. Tech Coloproctol. 2000;4:143–9.
51. Elsebae MM. A study of fecal incontinence in patients with chronic anal fissure: prospective, randomized, controlled trial of the extent of internal anal sphincter division during lateral sphincterotomy. World J Surg. 2007;31:2052–7.
52. Nyam DCNK, Wilson RG, Stewart KJ, et al. Island advancement flaps in the management of anal fissures. Br J Surg. 1995;38:69–71.
53. Leong AF, Seow-Choen F. Lateral sphincterotomy compared with anal advancement flap for chronic anal fissure. Dis Colon Rectum. 1995;38:69–71.
54. Singh M, Sharma A, Gardiner A, Duthie GS. Early results of a rotational flap to treat chronic anal fissures. Int J Colorectal Dis. 2005;20:339–42.
55. Giordano P, Gravante G, Grondona P, Ruggiero B, Porrett T, Lunniss PJ. Simple cutaneous advancement flap anoplasty for resistant chronic anal fissure: a prospective study. World J Surg. 2009;33:1058–63.
56. O'Kelly TJ, Brading A, Mortensen N. In vitro response of the human anal canal longitudinal muscle layer to cholinergic and adrenergic stimulation: evidence of sphincter specialization. Br J Surg. 1993;80:1337–41.
57. Rattan S. The internal anal sphincter: regulation of smooth muscle tone and relaxation. Neurogastroenterol Motil. 2005;17 Suppl 1:50–9.
58. Rattan S, Regan RF, Patel CA, De Godoy MA. Nitric oxide not carbon monoxide mediates nonadrenergic noncholinergic relaxation in the murine internal anal sphincter. Gastroenterology. 2005;129:1954–66.
59. Cook TA, Brading AF, Mortensen NJ. Differences in contractile properties of anorectal smooth muscle and the effects of calcium channel blockade. Br J Surg. 1999;86:70–5.
60. Chong PS, Bartolo DC. Hemorrhoids and fissure-in-ano. Gastroenterol Clin North Am. 2008;37:627–44.
61. O'Kelly TJ, Brading A, Mortensen N. Nerve mediated relaxation of the human internal anal sphincter: the role of nitric oxide. Gut. 1993;34:689–93.
62. Griffin N, Zimmerman DD, Briel JW, et al. Topical L-arginine gel lowers resting anal pressure: possible treatment for anal fissure. Dis Colon Rectum. 2002;45:1332–6.
63. O'Kelly TJ, Davies JR, Brading AF, Mortensen NJ. Distribution of nitric oxide synthase containing neurons in the rectal myenteric plexus and anal canal. Morphologic evidence that nitric oxide mediates the rectoanal inhibitory reflex. Dis Colon Rectum. 1994;37:350–7.
64. O'Kelly TJ, Davies J, Tam P, Mortensen NJM, Brading AFB. Abnormalities of nitric oxide producing neurones in Hirschsprung's disease; morphology and implications. J Pediatr Surg. 1994;29:294–9.
65. Ball C. The rectum, its diseases and developmental defects. London: Hodder and Stoughton; 1908.

66. Miles W. Observations upon internal piles. Surg Gynecol Obstet. 1919;29:497–506.
67. Abel AL. The pectin: the pectin band: pectenosis and pectenotomy. Lancet. 1932;i:714–8.
68. Eisenhammer S. The surgical correction of chronic anal (sphincteric) contracture. S Afr Med J. 1951;25: 486–9.
69. Rankin FW, Bargen JA, Buie LA, editors. The colon, rectum and anus. Philadelphia: WB Saunders; 1932. p. 584–93.
70. Blaisdell PC. Pathogenesis of anal fissure and implications as to treatment. Surg Gynecol Obstet. 1937;65: 672–7.
71. Milligan ETC. The surgical anatomy and disorders of the perianal space. Proc R Soc Med. 1942;36: 365–78.
72. Gabriel WB. The principles and practice of rectal surgery. 3rd ed. London: HK Lewis; 1945. p. 129–45.
73. Brown AC, Sumfest JM, Romadowski JV. Histopathology of the internal anal sphincter in chronic anal fissure. Dis Colon Rectum. 1989;32: 680–3.
74. Bo K, Lilleas F, Talseth T, et al. Dynamic MRI of the pelvic floor muscles in an upright sitting position. Neurourol Urodyn. 2001;20:167–74.
75. Schmeiser G, Putz R. The anatomy and function of the pelvic floor. Radiologe. 2000;40:429–36.
76. Shafik A. A new concept of the anal sphincter mechanism and physiology of defecation. IX. Single loop continence: a new theory of the mechanism of anal continence. Dis Colon Rectum. 1980;23:37–46.
77. Li D, Guo M. Morphology of the levator ani muscle. Dis Colon Rectum. 2007;50:1831–9.
78. Poh A, Tan KY, Seow-Choen F. Innovations in chronic anal fissure treatment: a systematic review. World J Gastrointest Surg. 2010;2:231–41.
79. Parks AG, Porter NH, Hardcastle J. The syndrome of the descending perineum. Proc R Soc Med. 1966;59:477–82.
80. Tan KY, Seow-Choen F, Hai CH, Thye GK. Posterior perineal support as treatment for anal fissures–preliminary results with a new toilet seat device. Tech Coloproctol. 2009;13:11–5.
81. D'Hoore A, Penninckx F. Obstructed defecation. Colorectal Dis. 2003;5:280–7.
82. Keighley MRB, Williams NS. Surgery of the anus, rectum and colon. London: WB Saunders; 1993.
83. McDonald P, Driscoll AM, Nicholls RJ. The anal dilator in the conservative management of acute anal fissures. Br J Surg. 1983;70:25–6.
84. Lock MR, Thompson JP. Fissure in ano: the initial management and prognosis. Br J Surg. 1977;64: 355–8.
85. Abramowitz L, Sonhani I, Benifla JL, et al. Anal fissure and thrombosed external hemorrhoids before and after delivery. Dis Colon Rectum. 2002;45: 650–5.
86. Hancock BD. The internal sphincter and anal fissure. Br J Surg. 1977;64:92–5.
87. Lin JK. Anal manometric studies in hemorrhoids and anal fissures. Dis Colon Rectum. 1989;32:839–42.
88. Lund JN. Nitric oxide deficiency in the internal anal sphincter of patients with chronic anal fissure. Int J Colorectal Dis. 2006;21:673–5.
89. Gibbons CP, Read NW. Anal hypertonia in fissures: cause or effect? Br J Surg. 1986;73:443–5.
90. Lund JN, Scholefield JH. Internal sphincter spasm in anal fissure. Br J Surg. 1997;84:1723–4.
91. Jonas M, Scholefield JH. Anal fissure. Gastroenterol Clin North Am. 2001;30:167–81.
92. Mearin F, Mourelle M, Guarner F, et al. Patients with achalasia lack nitric oxide synthase in the gastro-oesophageal junction. Eur J Clin Invest. 1993;23: 724–8.
93. Klosterhalfen B, Vogel P, Rixen H, Mittermayer C. Topography of the inferior rectal artery: a possible cause of chronic, primary anal fissure. Dis Colon Rectum. 1989;32:43–52.
94. Schouten WR, Briel JW, Auwerda JJ. Relationship between anal pressure and anodermal blood flow. The vascular pathogenesis of anal fissures. Dis Colon Rectum. 1994;37:664–9.

Chronic Anal Fissure

16

Siraj Rajaratnam and Ian Lindsey

Introduction

An anal fissure is a tear in the mucocutaneous lining of the anal canal. The true incidence of anal fissure is not known as it is likely that many people with it do not seek medical attention. It is certainly a very common condition.

Fissures can be classified as acute when symptoms have been present for less than 6 weeks or chronic if persisting for longer periods. Chronic fissures can be further divided into typical and atypical. The majority are typical fissures, which are primary in nature, and are the focus of this chapter. Atypical fissures are much rarer and are secondary to other conditions such as Crohn's disease, HIV, immunosuppression, malignancy, syphilis, tuberculosis or drug-induced, e.g. nicorandil.

Chronic anal fissures are usually associated with internal anal sphincter spasm, relief of which is central to promoting fissure healing. The treatment of chronic anal fissure has undergone a transformation in recent years from surgical to medical, both approaches sharing the common goal of reducing this spasm. This chapter examines some of the evidence for concern about traditional surgical treatment of chronic fissures and proposes a sphincter-saving approach to the patient with a chronic anal fissure.

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Clinical Assessment and Investigation

The classic symptoms of anal fissure are anal pain and bleeding. The pain is usually severe and is precipitated by defaecation. It may last for several minutes to a few hours afterwards. An atypical fissure should be suspected if a fissure is painless. The bleeding is usually described as bright red blood noted on the toilet paper after wiping following a bowel motion, without dripping into the bowl and not mixed with the stool. Constipation and straining at stool are often present, which exacerbate the symptoms. Specific questions should be asked with a view to excluding conditions associated with atypical fissures.

Clinical examination will usually reveal a linear split in the anoderm, which may be visible simply with separation of the buttocks. Anal hypertonia is noted on digital examination which is often painful. In acute fissures the edges of the defect are clean. Chronic fissures are ischaemic ulcers characterised by fibrotic rolled edges and a paucity of granulation tissue at the base where the transverse fibres of the internal sphincter are exposed. Other characteristic findings of chronic fissures are a sentinel tag at the apex of the fissure externally and a hypertrophied anal papilla at the internal apex. The majority of

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typical anal fissures occur in the posterior midline, with the remainder in the anterior midline. If the fissure is elsewhere in the anal canal, causes of atypical fissure should be considered.

Digital examination and proctoscopy are sufficient to make the diagnosis but may not be possible due to anal pain and spasm. If the history is classical with no atypical features, a trial of empirical medical management is appropriate. If there is any concern about diagnosis or cause, examination under anaesthesia and/or flexible endoscopy should be considered.

Even though digital rectal examination may be unreliable in differentiating a hypertonic from a normotonic anus [1], anal manometry is usually not required prior to medical treatment of anal fissure. Manometry is of use to detect a normotonic or hypotonic anus in those patients who fail initial medical treatment, prior to considering further treatments aimed at reducing sphincter spasm. Similarly, endoanal ultrasonography is not necessary as a routine investigation but has a role in excluding an occult sphincter injury prior to considering surgical sphincterotomy.

Pathophysiology

The exact pathophysiology of typical anal fissures is uncertain. Acute fissures are probably caused by trauma from passage of hard stool, childbirth, anorectal intercourse, or instrumentation of the anus [2]. The progression from acute to chronic fissure and the posterior location of most fissures can be explained by internal anal sphincter hypertonia and relative ischaemia of the posterior anal canal. These factors, alone or in combination, are the most popular hypotheses and provide the basis for current management of chronic anal fissure.

Mean resting anal pressures are higher in patients with posterior midline chronic anal fissures when compared with controls [3]. There is some disagreement as to whether the anal spasm leads to fissure formation or if it occurs secondarily as a result of the severe pain caused by the fissure. In either case, it appears that once an anal fissure is present, hypertonia of the anal sphincter may cause poor healing and persistence of the fissure. This occurs by way of reduced perfusion pressure of the mucocutaneous lining of the anus leading to the formation of an ischaemic ulcer, i.e. a chronic anal fissure [2]. Current management of anal fissure is aimed at reducing the anal pressure, thus improving mucocutaneous blood flow.

There are, however, a significant proportion of patients with typical chronic anal fissure who have normal anal pressures. Manometric studies have suggested that this may include around 50 % of patients [1, 3]. This would suggest that there are other factors involved. Arteriolar branches are fewer and capillaries less dense in the posterior anal canal possibly leading to a relative ischaemia in this area and explaining the predilection for anal fissures to occur in the posterior midline [2]. Anti-endothelial cell antibodies (AECA) may play a role. A small study found AECA in the peripheral blood of 90 % of patients with chronic anal fissure, whereas only 30 % of those with haemorrhoids and none of the asymptomatic controls were AECA positive [4].

We believe that there are three main pathophysiologic factors which lead to persistence of a chronic anal fissure: sphincter hypertonia (spasm), chronic fissure fibrosis and anorectal dysfunction. One or all of these may play a role in any individual patient to varying degrees. Determining which of these is the predominant factor is key in deciding on appropriate management for each individual patient.

Management of Chronic Anal Fissure

Surgery has been the mainstay of treatment for chronic anal fissure for many years, but concern over rates of permanent incontinence after surgery has driven the development of pharmacological therapies.

Traditional Surgical Therapies and the Risk of Incontinence

Traditional surgical therapies for chronic anal fissure include manual dilatation of the anus and lateral internal sphincterotomy.

Manual Dilatation of the Anus

Manual dilatation of the anal canal has been part of clinical practice for almost two centuries and has remained the primary treatment for fissure until as recently as the last two decades. Its goal is to reduce anal pressure by manual stretching of the internal sphincter. Under general anaesthesia, two, four, six or eight fingers are inserted into the anus and sustained lateral distraction exerted on the sphincters. This procedure causes uncontrolled disruption of the anal sphincters and thus a high risk of flatus and faecal incontinence. Injury to the internal sphincter, external sphincter or both can be demonstrated with endoanal ultrasonography.

The risk of incontinence after manual dilatation of the anus has been documented by both prospective and retrospective studies. There are problems with standardising interpretation and gradation of incontinence symptoms, but a 20–25 % risk is a reasonable estimate. The Cochrane meta-analysis updated in January 2010 supports this, giving an odds ratio of 4.03 (95 % CI 2.04–7.46) for flatus incontinence or faecal seepage after dilatation when compared with sphincterotomy [5].

Renzi et al. have reported a potentially safer form of anal dilatation in a randomised trial which involved use of a pneumatic balloon to control the amount of stretch applied to the anal canal [6]. A 40 mm diameter balloon was inflated to 20 psi pressure for 6 min under local anaesthesia with conscious sedation. Twenty-four patients underwent the procedure in a randomised trial comparing it with lateral sphincterotomy. Post-procedure ultrasonography showed no significant disruption of sphincters in the balloon dilatation group, and none of these patients were incontinent, whereas 16 % in the sphincterotomy group were incontinent. Fissure healing was not statistically different with 83.3 % of fissures healed with balloon dilatation compared to 92 % in the sphincterotomy group [6]. The recent Cochrane meta-analysis, however, found that dilatation was less effective than sphincterotomy in terms of fissure healing with an odds ratio of 1.55 (95 % CI 0.85–2.86) for persistence of fissure, favouring sphincterotomy [5].

Lateral Internal Sphincterotomy

Sphincterotomy has also been practised since the early nineteenth century, but it was not until the 1950s when Eisenhammer properly described the anatomy of the anal canal that partial division of the internal anal sphincter became a popular treatment for anal fissure [2]. Though effective, Eisenhammer's posterior midline internal sphincterotomy was complicated by significant post-operative pain, prolonged healing times and a residual "keyhole" deformity resulting in significant disturbance to continence [2]. Notaras subsequently reported lateral internal sphincterotomy which yielded similar fissure healing rates with significantly fewer complications. Lateral internal sphincterotomy remains the most effective single treatment for anal fissure with healing rates of over 90 %, but incontinence rates range from 0 to 30 % [5].

Lateral internal sphincterotomy can be performed in the lithotomy or prone jackknife position under general, regional or local anaesthesia. Bowel preparation and antibiotic prophylaxis are not necessary. After preparing and draping the perianal region, an Eisenhammer bivalve speculum or similar anal retractor is inserted into the anus and the intersphincteric groove palpated. A short circumferential incision is made laterally at either 3 or 9 o'clock and the internal sphincter identified. Submucosal and intersphincteric planes are developed to isolate the internal sphincter which may then be divided under direct vision. The caudal part of the internal sphincter is divided for a variable distance cranially, usually to the dentate line. The wound may be closed with fine absorbable interrupted sutures or may be left open. Routine post-operative care typically includes a stool-bulking agent and non-opiate analgesia [7].

The theoretical advantage of sphincterotomy over manual dilatation is based on the premise that the former involves a more controlled, partial division of the internal sphincter, with a consequent limited reduction in resting anal pressure of 25–35 % [8]. Yet permanent incontinence rates are still as high as 30 % [5]. There are many possible explanations for this. Surgical sphincterotomy is perhaps not as controlled and reproducible as one might

intend. The internal sphincter may be inadvertently divided along its entire length. Female patients, with relatively shorter sphincters, are at greater risk of this. The external anal sphincter may also be inadvertently divided, and pre-existing occult external sphincter defects (e.g. obstetric) that were previously compensated for may now become clinically apparent. Not all chronic fissures are associated with raised resting pressure. A manometric study of 40 patients with chronic anal fissure found that 19 % of men and 42 % of women had low or low-normal resting pressures and that surgeons were poor at identifying this group with digital examination alone [1]. Performing sphincterotomy in this group of patients is not only illogical but potentially risks incontinence with a surgical reduction in resting pressure of 25 %.

Techniques to Make Sphincterotomy Safer

A number of technical variations have been proposed with a view to reducing risk of incontinence whilst maintaining high healing rates. The traditional description of open lateral sphincterotomy involves dividing the full thickness of internal sphincter up to the level of the dentate line. In an attempt to reduce incontinence rates, various methods of tailoring the extent of sphincter division have been reported [9–12]. The most commonly reported variation is to divide the internal sphincter to the level of the internal apex of the fissure, stopping short of the dentate line. Littlejohn and Newstead published a series of 287 patients undergoing tailored sphincterotomy to the fissure apex. Of these, 4 (1.4 %) suffered flatus incontinence, 1 (0.35 %) minor soiling and 2 (0.7 %) urgency. There were no reports of faecal incontinence. Fissure healing rate was 99.65 % [9]. A subsequent randomised controlled trial has shown a significant reduction in post-operative incontinence with sphincterotomy to the fissure apex compared with dividing to the dentate line [10]. Elsebae reported incontinence rates of 2.17 % versus 10.86 % ($p=0.039$) but reported a statistically significant increase in post-operative days to relief of pain (3.42 vs 2.07 days) and a trend to prolonged healing time and increased treatment failure (4.35 % vs 0 %) in the tailored sphincterotomy group [10].

Closed or subcutaneous sphincterotomy is a slight modification of the open procedure which leaves a tiny perianal wound. With an Eisenhammer retractor or finger in the anal canal, a scalpel is inserted in a lateral position at the intersphincteric groove and advanced cephalad in either the submucosal or intersphincteric planes. The blade is then turned laterally (if submucosal) or medially (if intersphincteric) to divide the internal sphincter whilst feeling the characteristic "give" as the tension in the muscle is released [7]. The blade is removed, haemostasis achieved with pressure and the wound left open. A large retrospective series from Minnesota compared open ($n=521$) and closed ($n=343$) sphincterotomy [11]. At median follow-up of 3 years, there was no difference between the two groups in fissure healing rates, recurrence and need for reoperation. There was, however, increased rates of flatus incontinence (30.3 vs 23.6 %, $p=0.062$), faecal leakage (26.7 vs 16.1 %, $p<0.001$) and accidental bowel movements (11.8 vs 3.1 %, $p<0.001$) in the open sphincterotomy group. The authors recommend closed sphincterotomy based on these findings [11]. This recommendation, however, is not supported by the Cochrane meta-analysis of 5 randomised trials (299 patients) comparing open and closed sphincterotomy. The meta-analysis found no significant difference in fissure persistence or flatus incontinence but did not include incidence of faecal incontinence [5].

Rosa et al. described another method of tailoring the length of sphincterotomy [12]. In their series of 388 patients, pre-operative anal manometry was used to determine the degree of sphincter hypertonia (mild, moderate, severe). 20, 40 or 60 % of the internal sphincter was then divided corresponding to the degree of hypertonia. Of the 261 patients with available follow-up data, 96.2 % were cured at median follow-up 8 months, and just one patient complained of flatus incontinence [12].

A Conservative, Sphincter-Sparing Management Approach

Due to the risk of permanent continence disturbance with traditional surgical therapies, medical alternatives, such as topical creams and

botulinum toxin injection, have been developed with the aim of reducing mean resting anal pressure for a temporary period, allowing the fissure to heal, without any long-term risk of incontinence. Other surgical procedures, such as fissurectomy and anal advancement flap, have also been introduced.

Figure 16.1 provides a management algorithm which aims to make sense of the multitude of options now available to the surgeon. We developed this algorithm to pragmatically identify the likely pathophysiologic process (sphincter spasm, chronic fibrosis or anorectal dysfunction) that is causing the fissure to persist and targeting treatment accordingly with the intention of healing the fissure without risking permanent continence disturbance.

The various components of the algorithm are explained below.

Medical Management of Sphincter Spasm

Topical Creams: Nitric Oxide Donors and Calcium Channel Blockers

Primary therapy for chronic anal fissure consists of a topical agent combined with a stool softener.

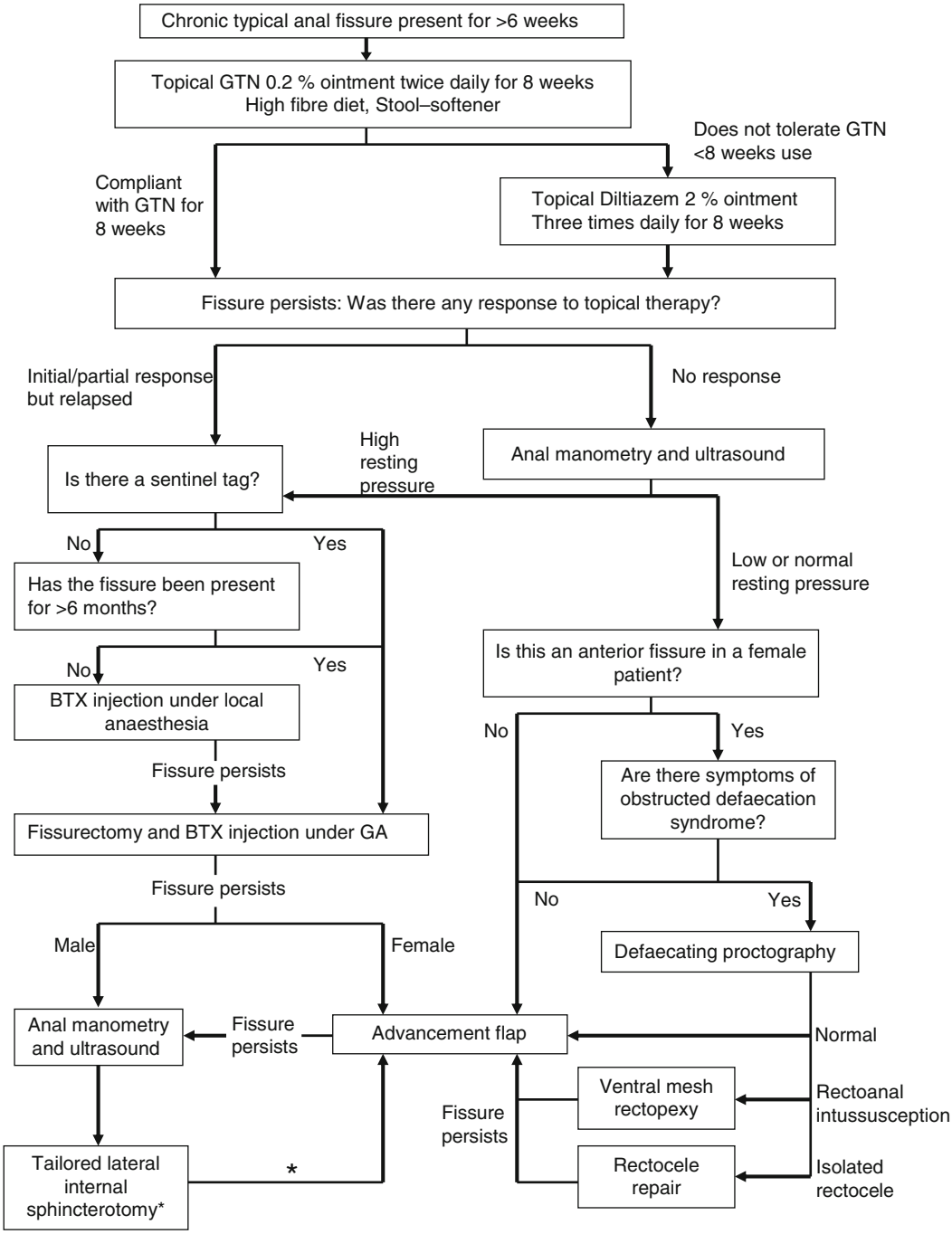
The first pharmacological treatments developed were the nitric oxide donors. These cause vasodilation and smooth muscle relaxation and are long established in the management of coronary artery disease. Topical glyceryl trinitrate (GTN) causes relaxation of the internal sphincter [13] and is the most studied of all medical therapies for anal fissure. The usual course is 0.2 %, applied directly to the anus two or three times daily, for 8 weeks. The Cochrane meta-analysis published in 2008 included 15 randomised trials (1,190 patients) and reported an overall fissure healing rate of 48.6 % for GTN paste versus 37 % for placebo [13]. This was statistically significant. The principal side effect of GTN is headache which may occur in up to 50 %.

Direct comparison of trials is complicated by the different dosages of GTN used, the criteria used for distinguishing chronic and acute fissures and the duration of symptoms prior to treatment. However, GTN is still considered first-line therapy in many centres as it is easily available and its safety is undisputed. However, as half of patients treated with GTN will not respond [13], the downside for the patient is 6–8 weeks of persistent symptoms and possibly headache with the ultimate need for second-line treatment. There are few data on whether these patients should be scheduled for surgery after treatment failure or whether further medical therapy confers any benefit.

Despite its frequent use, the role of topical GTN in chronic anal fissure remains uncertain. Although it showed early promise in healing fissure, initial enthusiasm has been tempered somewhat by concerns over medium-term relapse, headache and tachyphylaxis. As a topical preparation, there are inevitable difficulties with regulating dose, and non-compliance may also be common.

There are probably additional explanations for the shortfall in healing rates with GTN. Pitt et al. followed 64 patients treated with GTN with the aim of identifying independent risk factors for treatment failure. Failure to heal was more likely in the presence of a sentinel tag ($p<0.035$) and in fissures present for more than 6 months ($p<0.05$) [14]. This suggests an inability of medical therapy to heal fissures at the truly chronically established end of the spectrum. There is also a subgroup of patients with chronic fissure who fail to exhibit reductions in resting pressure in response to topical GTN, and healing seems unlikely in these patients [15]. Furthermore, patients with low or normal resting anal pressures may also fail GTN therapy as sphincter spasm is probably not the principal underlying reason for persistence of the fissure in these patients.

Calcium channel blockers, such as diltiazem and nifedipine, lower resting anal pressure by causing smooth muscle relaxation. As with GTN, there is also a vasodilator effect, hence the established role of these drugs as antihypertensives. For anal fissure, topical calcium channel blockers are applied for 6–8 weeks. Four trials with heterogenous methodologies, comparing calcium channel blockers with GTN, were included in a Cochrane Review [13]. These studies are limited by small sample size or short



* Relative contraindications to sphincterotomy must be considered i.e. female patients, pre–existing continence disturbance, older patients, low resting pressure, sphincter defect

Fig. 16.1 Algorithm for management of chronic anal fissure

follow-up, but there was no significant difference in healing rates between GTN and calcium channel blockers, whereas side effects such as headache are more common with GTN. In the United Kingdom, anal fissure is at present an unlicensed indication for topical diltiazem although it may be used in patients who have not responded to topical nitrates.

A study of 64 patients comparing topical 0.5 % nifedipine with lateral sphincterotomy reported healing rates of 96.7 % at 8 weeks and 93.3 % at 19 months in the nifedipine group, which was not significantly less than the 100 % in the lateral sphincterotomy group [16]. Overall side effects were higher with nifedipine (headache 15.6 % vs 0 %; flushing 15.6 % vs 0 %; anal irritation 18.7 % vs 6.2 %), but 12.5 % of patients in the sphincterotomy group developed permanent flatus incontinence as contrasted by 0 % in the nifedipine group [16].

Whilst the efficacy of topical medical therapies remains in question, and the side effects are well documented, there are no concerns regarding long-term safety. Therefore, we believe that primary therapy for chronic anal fissure should be with a topical agent. We use GTN as the first-line therapy, reserving diltiazem for those who are intolerant to GTN.

A course of topical therapy is also useful in identifying those patients who may have low or normal pressure fissures and would be less likely to respond to escalation of treatments aimed at reducing resting pressure (e.g. botulinum toxin and internal sphincterotomy). It is not practical to perform anal manometry in every patient with chronic anal fissure, so response to topical therapy is used to select those patients in which manometry is likely to be helpful. For example, a patient who is fully compliant with an 8-week course of topical therapy, yet does not respond at all, should go on to have anal manometry studies for measurement of sphincter pressures, whereas a patient who partially responds, or responds and relapses, probably has a high pressure fissure. Reasons for persistence or recurrence of these fissures may include chronic fibrosis, poor compliance or the reduction in resting pressure being inadequate or not sustained for long enough.

Botulinum Toxin A

Botulinum toxin A (BTX), a neurotoxin produced by *Clostridium botulinum*, is established in cosmetic medicine and for hypertonic skeletal muscle disorders. In skeletal muscle it inhibits the release of acetylcholine at the neuromuscular junction, but its action on the smooth muscle of the internal sphincter is less clear. A study using a porcine model found that BTX reduces myogenic tone and contractile response to sympathetic stimulation, either directly on smooth muscle or indirectly on the nerves, perhaps through acetylcholine [17]. When injected into muscle, BTX produces a paralysis of predictable duration (2–4 months). The aim of its use in treating chronic anal fissure is to cause a sustained reduction in resting anal pressure but for a temporary period to allow fissure healing without compromising long-term continence.

BTX is supplied in powder form which requires reconstitution with normal saline. Injection may be done in the outpatient clinic or under general anaesthesia. A total of 20–40 units of Botox™ or 100–200 units of Dysport™ is injected into the internal sphincter or the intersphincteric space on either side of the anterior or posterior midline with a fine-bore (27-G) needle. A number of alternative methods of injection, including unilateral injection, more than two injection sites and injection into the external sphincter, have been reported.

A placebo-controlled double-blind randomised trial of 30 patients published in 1998 demonstrated BTX to be considerably more effective than saline injection [18]. Patients in the intervention group were treated with a total of 20 units of Botox™ injected into the internal sphincter adjacent to the fissure on both sides. Fissure healing rates were 53 and 73 % at 1 and 2 months, respectively, in the BTX group. In the control group, only 13 % were healed at 2 months ($p=0.003$). No continence disturbances occurred in the BTX group. One patient in the control group suffered temporary flatus incontinence [18].

As well as placebo, BTX has been compared with GTN and lateral internal sphincterotomy in randomised trials presented in a meta-analysis by the Cochrane Collaboration [13]. With the exclusion

of three outlying studies, the Cochrane Review found BTX to be superior to placebo, equivalent to GTN and inferior to surgical sphincterotomy, with an overall healing rate with BTX of 65 % [13].

For several reasons, BTX injection has distinct advantages over other current medical therapies. BTX is administered by one-off injection, which seems to be well tolerated, thus eliminating non-compliance issues. A single injection into the internal sphincter produces a reduction in resting pressure of a similar order to that obtained with GTN. However, BTX produces a constant reduction in resting pressure sustained over a 2–3-month period which should translate to improved healing. Side effects such as temporary minor incontinence, urgency and perianal haematoma are infrequent and not sustained. Its major disadvantage is its cost, though this can be set against the cost of surgical therapy.

We use BTX in patients who have partially responded to topical agents, in those who have responded and relapsed and in those who did not respond but have high resting pressure on anal manometry (Fig. 16.1). In the absence of a sentinel tag, and in chronic fissures present for less than 6 months, BTX is injected under local anaesthetic anal block (10 mL 0.5 % bupivacaine and 10 mL 1 % lignocaine) in an outpatient setting. If there are signs of chronic fibrosis, BTX injection is combined with fissurectomy under general anaesthesia (see below).

Management of Chronic Fissure Fibrosis

Pitt et al. identified the presence of a sentinel tag and persistence for more than 6 months as independent risk factors for poor healing [14]. Chronic fibrosis cannot be treated by medical therapy, hence the failure to heal despite adequate treatment of the internal sphincter spasm.

Fissurectomy

Fissurectomy is excision of the fissure complex (hypertrophied anal papilla, sentinel tag, fibrotic scar tissue) down to but not including the internal sphincter, leaving a fresh surgical wound which may be advantageous for healing [19].

Fissurectomy alone has been suggested by surgeons who believe that sphincter hypertonia is a consequence of fissure rather than the cause [19]. A series of 118 patients undergoing fissurectomy alone resulted in similar healing and incontinence rates to internal sphincterotomy [19].

Combining medical management of sphincter spasm with surgical excision of the fibrotic tissue (fissurectomy) has been shown to be of benefit in a case series of 30 patients [20]. All had previously failed medical management (19 GTN, 11 GTN and BTX) and were treated with combined BTX and fissurectomy. The authors reported a fissure healing rate of 93 % at median 16.4 weeks follow-up. The two patients whose fissures did not heal had significant symptomatic improvement such that no further management was required. Two patients (7 %) reported flatus incontinence which had resolved in both cases by 6 weeks, and there were no cases of faecal incontinence [20].

If primary treatment with topical GTN or diltiazem fails, we combine BTX injection and fissurectomy under general anaesthesia in patients with sentinel tag, fissure present for more than 6 months and in those with failure to heal after outpatient BTX administration (Fig. 16.1).

Advancement Flap

The use of an advancement flap of perianal skin to cover the anal fissure was assessed in a randomised trial of 40 patients comparing it with lateral sphincterotomy [21]. Healing rates were 85 % vs 100 % (p = NS), and no patient in either group was incontinent. Median operating time was twice as long in the flap group (10 vs 5 min). Advancement flap appears to be a safe and effective treatment, certainly for low and normal pressure fissures and in those with recalcitrant high pressure fissures who may be at particular risk of incontinence with sphincterotomy, e.g. females, older age group, pre-existing continence disturbance, and sphincter defect on ultrasound (Fig. 16.1).

Management of Underlying Anorectal Dysfunction

Low or normal pressure fissures, particularly an anterior fissure in a female patient, appear to have a pathophysiology that differs to the

conventional theory of sphincter hypertonia, ischaemia and chronic fibrosis. Anorectal dysfunction appears to play a role in these patients. Ellis demonstrated in 54 women with an anterior fissure and a rectocele 100 % healing rates with levatorplasty compared with 96 % in those randomised to sphincterotomy [22]. Resting pressures did not change in the levatorplasty group but were significantly reduced in the sphincterotomy group. Further evidence for the role of anorectal dysfunction is a series of 11 patients with chronic anal fissures resistant to GTN and second-line diltiazem or botulinum toxin. Nine patients complained of symptoms of obstructed defaecation syndrome. Five of these patients had rectoanal intussusceptions demonstrated on defaecating proctography [23]. In another case series, five patients with chronic recalcitrant anal fissure and rectoanal intussusception on defaecating proctography (three also had a rectocele) underwent stapled transanal rectal resection (STARR). All of the fissures healed [24]. Another small series ($n = 5$) of patients with chronic anal fissures were treated with a temporary period of sacral neuromodulation, and all fissures were healed by 3 weeks [25].

In patients with a low or normal pressure anterior fissure, we believe that diagnosis and treatment of any underlying anorectal dysfunction are paramount to enable long-term healing of the fissure (Fig. 16.1).

The Role of Lateral Internal Sphincterotomy

There is no longer a role for manual dilatation of the anus in the modern management of chronic anal fissure. In highly selected patients in whom a sphincter-sparing approach has failed, tailored lateral internal sphincterotomy may still be indicated.

Anorectal manometric and ultrasonographic evaluation prior to surgery allow a more accurate assessment of risk and better informed consent. Some of these patients may indeed wish to avoid internal sphincterotomy altogether and persist with an alternative medical therapy or even put up with their current symptoms.

Conclusion

Chronic anal fissure is a common problem. Its pathophysiology may be multifactorial and include internal anal sphincter spasm and ischaemia, chronic fissure fibrosis and anorectal dysfunction.

The approach to primary therapy of chronic anal fissure, be it surgical or medical, remains a matter of philosophy. Internal sphincterotomy remains a good operation at bringing rapid pain relief with a high degree of patient satisfaction. However, concern remains about the lack of standardisation of the procedure, exposing some patients to a risk of permanent disturbances in anal continence, particularly in inexperienced hands. These concerns are not shared with a medical approach.

The emergence of medical therapies has certainly refocused attention on the efficacy and safety of internal sphincterotomy in comparison. Whilst undoubtedly effective and reasonably safe when a tailored sphincterotomy is performed by an experienced surgeon, we believe the key to making internal sphincterotomy safer is to minimise the size of the cohort exposed to its inherent long-term risk by using a conservative, sphincter-sparing approach and reserving sphincterotomy for patients who have failed medical therapy and are assessed and stratified to a low incontinence-risk category.

References

1. Jones OM, Ramalingam T, Lindsey I, Cunningham C, George BD, Mortensen NJ. Digital assessment of internal sphincter spasm in chronic anal fissure is unreliable. Dis Colon Rectum. 2005;48:349–52.
2. Lund JN, Scholefield JH. Aetiology and treatment of anal fissure. Br J Surg. 1996;83(10):1335–44.
3. Bove A, Balzano A, Perrotti P, Antropoli C, Lombardi G, Pucciani F. Different anal pressure profiles in patients with anal fissure. Tech Coloproctol. 2004;8(3):151–6.
4. Maria G, Brisinda D, Ruggieri MP, Civello IM, Brisinda G. Identification of anti-endothelial cell antibodies in patients with chronic anal fissure. Surgery. 1999;126(3):535–40.
5. Nelson R. Operative procedures for fissure in ano. Cochrane Database Syst Rev. 2010. doi:10.1002/14651858.CD002199.pub3.

6. Renzi A, Izzo D, Di Sarno G, Talento P, Torelli F, et al. Clinical, manometric, and ultrasonographic results of pneumatic balloon dilatation vs. Lateral internal sphincterotomy for chronic anal fissure: a prospective, randomized, controlled trial. Dis Colon Rectum. 2007;51:121–7.
7. Gordon PH. Fissure-in-ano. In: Nivatvongs S, Gordon PH, editors. Principles and practice of surgery for the colon, rectum, and anus. 3rd ed. New York: Informa Healthcare; 2007.
8. Williams N, Scott NA, Irving MH. Effect of lateral sphincterotomy on internal anal sphincter function. Dis Colon Rectum. 1995;38:700–4.
9. Littlejohn DR, Newstead GL. Tailored lateral sphincterotomy for anal fissure. Dis Colon Rectum. 1997; 40(12):1439–42.
10. Elsebae MM. A study of fecal incontinence in patients with chronic anal fissure: prospective, randomized, controlled trial of the extent of internal anal sphincter division during lateral sphincterotomy. World J Surg. 2007;31(10):2052–7.
11. Garcia-Aguilar J, Belmonte C, Wong WD, Lowry AC, Madoff RD. Open vs. closed sphincterotomy for chronic anal fissure: long-term results. Dis Colon Rectum. 1996;39(4):440–3.
12. Rosa G, Lolli P, Piccinelli D, Mazzola F, Zugna C, et al. Calibrated lateral internal sphincterotomy for chronic anal fissure. Tech Coloproctol. 2005;9:127–32.
13. Nelson RL. Non surgical therapy for anal fissure. Cochrane Database Syst Rev. 2008. doi:10.1002/14651858.CD003431.pub2.
14. Pitt J, Williams S, Dawson PM. Reasons for failure of glyceryl trinitrate treatment of chronic fissure-in-ano: a multivariate analysis. Dis Colon Rectum. 2001; 44(6):864–7.
15. Lund J, Scholefield JH. Glyceryl trinitrate is an effective treatment for anal fissure. Dis Colon Rectum. 1997;40:468–70.
16. Katsinelos P, Papaziogas B, Koutelidakis I, Paroutoglou G, Dimiropoulos S, et al. Topical 0.5% nifedipine vs. lateral internal sphincterotomy for the treatment of chronic anal fissure: long-term follow-up. Int J Colorectal Dis. 2006;21(2):179–83.
17. Jones OM, Brading AF, Mortensen NJ. The mechanism of action of botulinum toxin on the internal anal sphincter. Br J Surg. 2004;91:224–8.
18. Maria G, Cassetta E, Gui D, Brisinda G, Bentivoglio AR, Albanese A. A comparison of botulinum toxin and saline for the treatment of chronic anal fissure. N Engl J Med. 1998;338(4):217–20.
19. Aigner F, Conrad F. Fissurectomy for treatment of chronic anal fissures. Dis Colon Rectum. 2008;51(7): 1163.
20. Lindsey I, Cunningham C, Jones OM, Francis C, Mortensen NJ. Fissurectomy-botulinum toxin: a novel sphincter-sparing procedure for medically resistant chronic anal fissure. Dis Colon Rectum. 2004;47(11):1947–52.
21. Leong AF, Seow-Choen F. Lateral sphincterotomy compared with anal advancement flap for chronic anal fissure. Dis Colon Rectum. 1995;38(1):69–71.
22. Ellis CN. Anterior levatorplasty for the treatment of chronic anal fissures in females with a rectocele: a randomised controlled trial. Dis Colon Rectum. 2004;47:1170–3.
23. Wijffels N, Collinson R, Cunningham C, Lindsey I. Rectoanal intussusception frequently underlies recalcitrant chronic anal fissures. Colorectal Dis. 2008;10 Suppl 2:15.
24. Clarke AD, Chand M, Tarver D, Nash GF, Lamparelli M. Stapled transanal rectal resection in the management of resistant anal fissures. Colorectal Dis. 2008;10 suppl 1:6.
25. Yakovlev A, Karasev SA, Dolgich OY. Sacral nerve stimulation: a novel treatment of chronic anal fissure. Dis Colon Rectum. 2011;54:324–7.

Anal Sepsis: Anatomy, Pathophysiology and Presentation

17

Stanley M. Goldberg and Patrick S. Tawadros

Introduction

Perianal sepsis is one of the most commonly observed anorectal lesions, leading to abscess formation and often fistula development. Despite its prevalence, it also remains one of the most challenging anorectal conditions to treat, often requiring multiple attempts and different management strategies. Fundamental to the adequate management of these conditions, however, is an understanding of anorectal anatomy and pathophysiology leading to abscess and fistula formation. Of interest, history is replete with ancient accounts of anorectal surgery, from ancient Egypt to Hippocrates, to John Ardene of England's "Treatise on Fistula, Hemorrhoids and Clyster" written in 1367. A turning point for anorectal surgery came in 1686 when the French King Louis XIV, who suffered from a fistula-in-ano, ordered his physician Philip to conduct clinical trials on the available methods of fistula treatment at that time. After 1 year of studies, the first "evidence-based" decision in anorectal surgery was made and the king submitted to a fistulotomy that ultimately cured him [1].

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Anatomy

A thorough discussion of anorectal sepsis cannot be accomplished without an understanding of the pertinent anatomy. While Chap. 2 was entirely dedicated to the anatomy of the anal canal, here we specifically review the pertinent aspects to the pathophysiology of anal sepsis.

Anal Canal

The anal canal is characterized at its mid-point by a transition from intestinal columnar epithelium proximally to squamous epithelium distally, also called anoderm (Fig. 17.1a). The transition is gradual over a distance of 6–12 mm in what is termed the transition zone [2]. Grossly, the change in colour from the red/purple proximal intestinal epithelium to the white distal squamous epithelium is undulated and referred to as the dentate line. As the anal canal is approximately 4 cm in length (variable depending on patient build), the dentate line is approximately 2 cm proximal to the anal verge.

The proximal aspect of the canal is defined by the anorectal ring, comprised of the proximal aspects of the internal and external sphincter muscles as well as the puborectalis muscle (Fig. 17.1b). This important ring forms the junction between the rectum and the anal canal and is critically important during the treatment of perianal sepsis as division of this ring would

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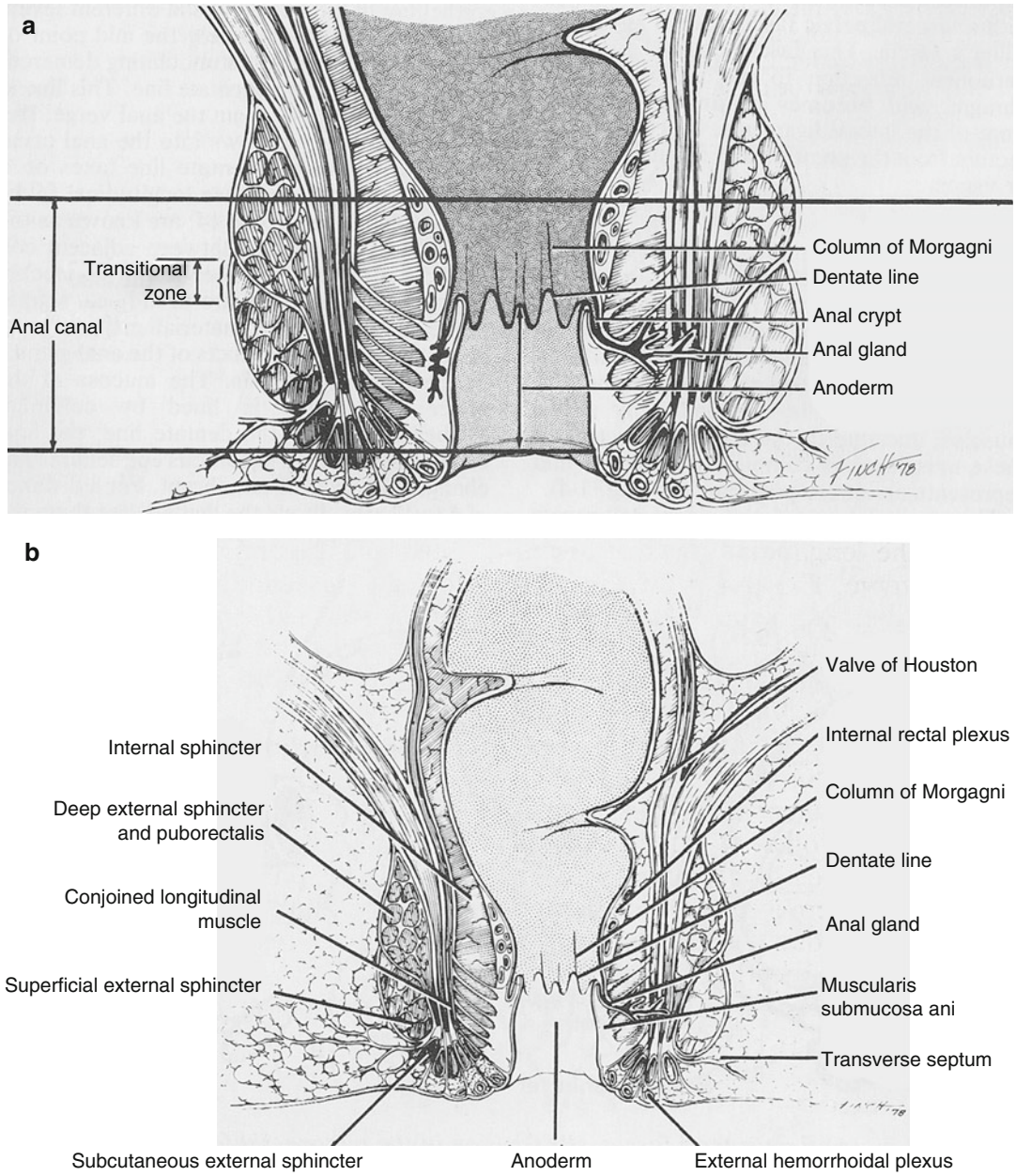

Fig. 17.1 (**a**) The anal canal [1]. (**b**) The anorectum [1]

inevitably affect continence. The distal aspect of the anal canal is the anal verge, defined as the area where anoderm becomes true skin characterized by pigmentation, hair, sweat glands and a ring of apocrine glands, which may contribute to the condition hidradenitis suppurativa.

Anal Glands

Because of the diminishing size calibre from rectum to anus, the undulated dentate line area of the anal canal is characterized by a pleated appearance, which is referred to as the columns of Morgagni. At the base of each column, at the

level of the dentate line, there is a small pocket or crypt. About half of these crypts communicate with the drainage ducts of anal glands that are located in the internal sphincter muscle area. These anal glands, which were first described by Hermann and Desfosses in 1844 [3], are located in the submucosal space, the internal sphincter muscle itself, and about half also communicate with the intersphincteric space, without involving the external sphincter muscle. Anal crypts are therefore of significant importance in the pathophysiology of anal sepsis as foreign material lodging itself in a crypt can obstruct the drainage path of anal glands and result in sepsis. The importance of this mechanism was first described by Parks in 1961 [4].

Perianal Spaces

The complex of muscles which surround and form the anal canal create a network of spaces that are of paramount importance in the management of perianal sepsis (Fig. 17.2a).

The *perianal space* is in the immediate area of the anal verge surrounding the anal canal. The lateral aspect is continuous with the subcutaneous fat of the buttocks, while medially, it extends into the anal canal to the dentate line. It includes the distal most aspect of the external sphincter muscle and communicates freely with the intersphincteric space. The space is itself divided by an array of radiating elastic septa that limit communication within the space and account for the severe pain produced by the accumulation of even a small amount of pus.

The *intersphincteric space* is the space between the internal and external sphincter muscles. Proximally it terminates in the rectal wall, while distally, it communicates with the perianal space. This is the space that is often first seeded by sepsis of cryptoglandular origin.

The *ischiorectal space or fossa* is delineated proximally by the levator ani muscle and extends distally to the skin of the perineum. Laterally, along the obturator foramen, it contains Alcock's canal which carries the internal pudendal vessels and nerve.

The *supralevator spaces* are located proximal to the levator muscle on either side of the rectum. They extend proximally or superiorly to the peritoneal reflection and are limited laterally by the pelvic wall. Sepsis in this area may come from upward extension of perianal sepsis or downward extension of pelvic sepsis. Differentiating the source of sepsis in this space is important for deciding the appropriate route of drainage.

The *postanal space* is directly posterior to the anal canal and altogether distal to the levator ani muscle (Fig. 17.2b). It is divided into *deep and superficial postanal space* based on whether it is deep or superficial to the anococcygeal ligament. This space is of significant importance for understanding the pathophysiology of bilaterally communicating ischiorectal space sepsis, also known as a horseshoe abscess.

Pathophysiology

Anorectal sepsis is a common condition that can occur in healthy, normal individuals. Since anorectal abscesses often lead to fistulae formation, it is reasonable to consider both on the same pathological continuum.

Aetiology

There are several reasons that can account for the development of anal sepsis. Most commonly, the cause is of cryptoglandular origin, whereby anal crypts are obstructed and the drainage path of anal gland secretions is blocked. As a result, pus accumulates and builds in a retrograde fashion often seeding the intersphincteric space [5]. Other notable causes of perianal sepsis include perianal trauma and anal foreign bodies, which can cause abscess formation by the same mechanism as inspissated faecal material in anal crypts. Iatrogenic causes such as surgical wounds, haemorrhoid banding or injection have also been reported to occasionally cause perianal sepsis. More recently, two reviews have been published on stapled hemorrhoidectomy as another potential cause of severe perianal, pelvic and even

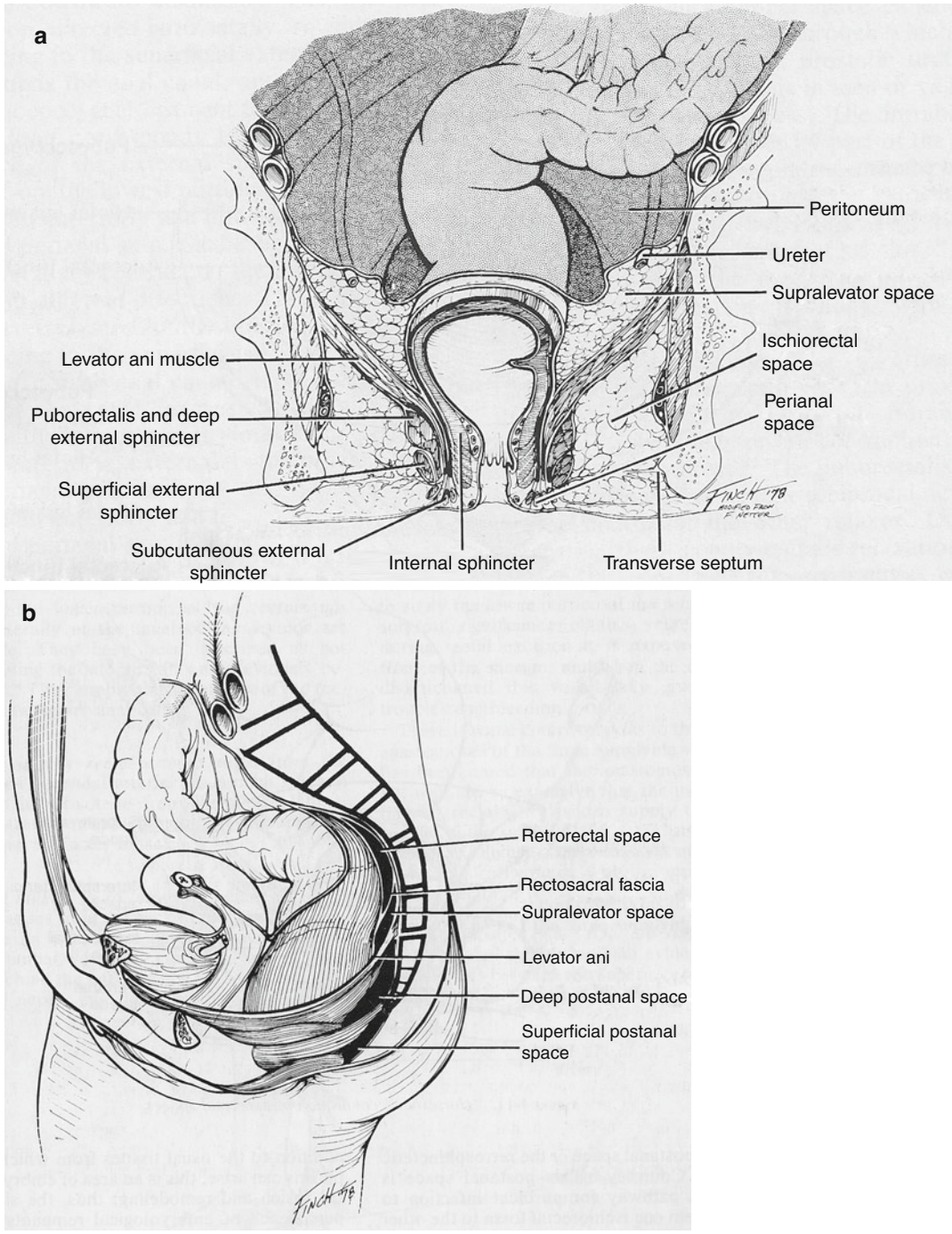

Fig. 17.2 (**a**) Perianal and perirectal spaces (frontal view) [1]. (**b**) Perianal and perirectal spaces (lateral view) [1]

intra-abdominal sepsis [6, 7]. Lastly, skin lesions such as hidradenitis suppurativa, localized pyoderma or infection of a sebaceous adenoma are also possible causes of perianal sepsis. Crohn's disease, while not itself a cause, is a strongly predisposing factor in the development of perianal abscesses, usually via the cryptoglandular mechanism.

Table 17.1 Aetiology of anorectal fistulas

Nonspecific (90 %)	Cryptoglandular
Specific (10 %)	Crohn's disease
	Chronic ulcerative colitis
	Trauma
	Foreign body
	Carcinoma (anal Ca, rectal Ca)
	Leukaemia/lymphoma
	Radiation
	Tuberculosis
	Lymphogranuloma granulosum
	Actinomycosis
	Others

Anal fistulas are also predominantly of cryptoglandular origin and will develop in approximately one-third of patients who undergo drainage of an anorectal abscess. A recent retrospective cohort study of 148 patients with anorectal abscesses showed a 37 % overall rate of fistula formation over a mean follow-up period of 38 months and a twofold increased risk of fistula formation in patients under the age of 40 [8]. While about 90 % of fistulas have a cryptoglandular aetiology, not all fistulas come from antecedent abscesses, and not all abscesses lead to fistula formation. Indeed a previous history of abscess may not be elicited in one-third of patients with anal fistula [9]. The remaining 10 % of anal fistulas have the following specific aetiologies (Table 17.1): anorectal disease, inflammatory bowel disease, infection, malignancy and trauma. It is important to distinguish the aetiology of an anal fistula in order to provide adequate treatment. Indeed, perianal fistulizing disease may be the initial presentation in 10 % of Crohn's patients, preceding the diagnosis by several years, and remains the only manifestation of disease in about 5 % of patients [10].

Bacteriology

Many studies have been performed looking at the bacteriology of pus in anorectal sepsis. A mixed flora of anaerobes, gut-derived gram negative organisms and some aerobic Staphylococcal species are found. Interesting studies have been performed to look at whether the bacteriology profile could be used to predict fistula formation. One conclusive such study was performed by Grace et al. in 1982 [11]. A prospective study of 165 patients presenting for incision and drainage of perianal abscess over 4 years had their pus sent for cultures. If a fistula was unapparent at the time of the first operation, another examination under anaesthesia was performed 10 days later for a second look, blinded to the culture results. The results were as follows: 54 % of the 114 patients who grew gut organisms were found to have a fistula, while none of the 34 patients who grew skin-derived organisms were found to have one. Other studies since then have confirmed this trend. The clinical implication from these studies is that all perianal abscesses should be cultured and only those that grow gut-derived organisms should be subjected to a second look for fistula formation if none was observed at the initial operation.

Spread of Infection

Perianal sepsis will spread using the path of least resistance. Commonly originating in the intersphincteric space, infection may spread distally into the perianal space, laterally through the external sphincter into the ischiorectal space or proximally along the intersphincteric space causing a supralevator abscess (Fig. 17.3). Pus can then originate from any of these locations to form complicated tracts that may result in fistula formation. Fistula trajectory can be very difficult to predict in patients with Crohn's disease; however, the majority of ordinary fistulas follow Goodsall's rule whereby tracts with external openings around the anterior half of the anus tend to have straight trajectories toward the nearest crypt, while those with an external opening around the posterior half of the anus will typically have a curved trajectory toward a posterior midline crypt (Fig. 17.4).

Horseshoe Abscess

Perianal sepsis may also spread to affect the contralateral side. This circumferential extension of

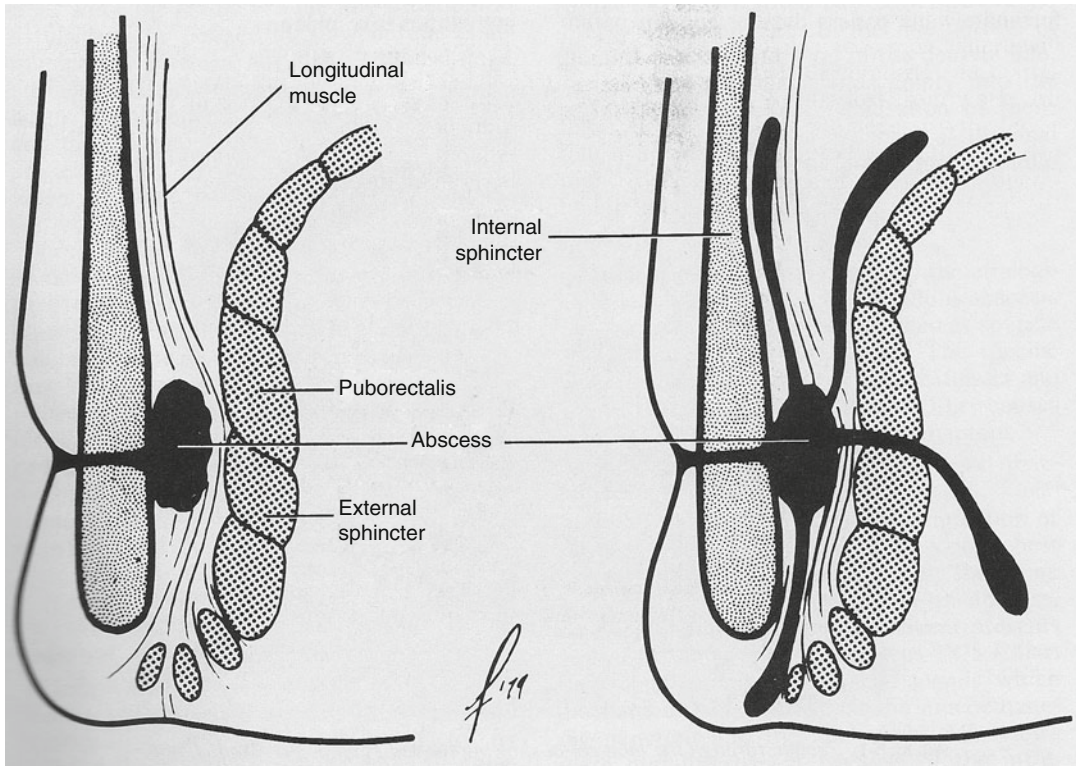

Fig. 17.3 Avenues of extension [1]

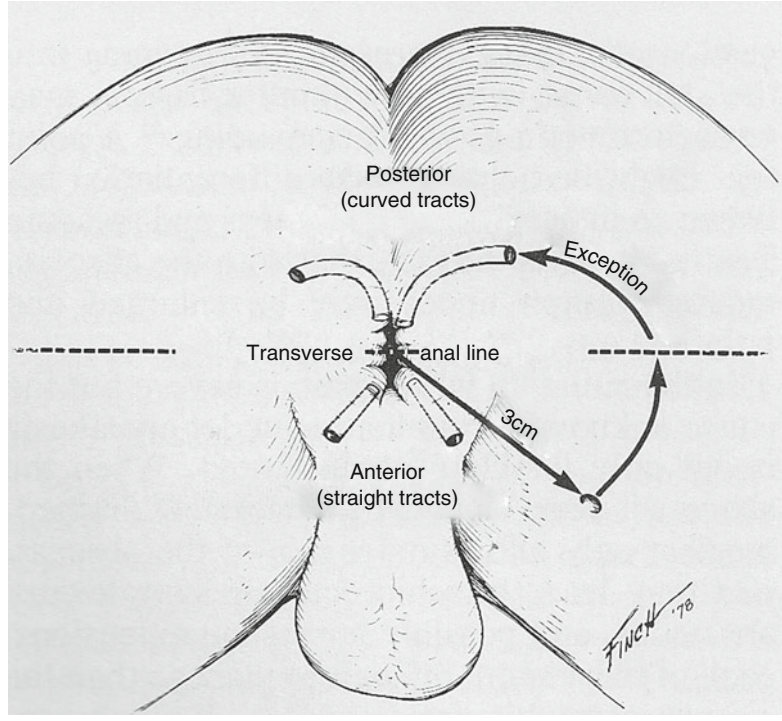

Fig. 17.4 Goodsall's rule [1]

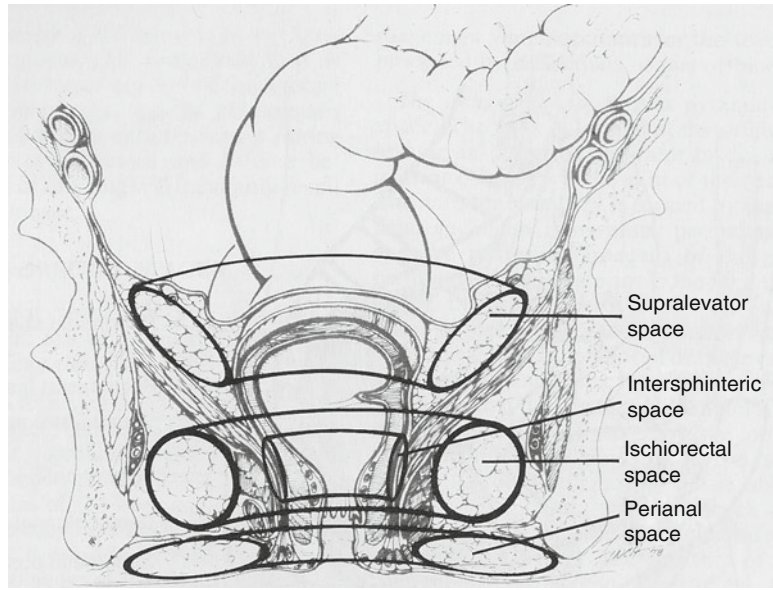

Fig. 17.5 Horseshoe connections of anorectal spaces [1]

infection can occur at any level within the intersphincteric, ischiorectal or supralevator spaces, resulting in a so-called horseshoe abscess (Fig. 17.5). The ischiorectal horseshoe abscess courses through the deep postanal space, while the supralevator horseshoe wraps around the rectum in the retrorectal space.

Presentation

Clinical Presentation

Acute perianal sepsis typically presents with new onset of pain and swelling in the affected area. The pain is aggravated by movement and defecation and sometimes even coughing or sneezing. Systemically, the pain is often accompanied by general malaise and fever. A clinical history may also reveal an antecedent bout of diarrhoea.

In the scenario of an intersphincteric abscess, the pain can often persist undiagnosed. The pain is described as throbbing in character through day and night, and cause extreme pain on defecation to the point of causing secondary faecal impaction. Occasionally, minor anal bleeding and/or purulent discharge may be observed.

In the chronic scenario, the patient will often describe a history of pain followed by spontaneous purulent discharge and subsequent temporary relief. When drainage is incomplete, the abscess cavity may re-accumulate and the internal opening may bleed from chronic granulation tissue.

On examination, the cardinal signs of inflammation are typically present and include redness, swelling, warmth and tenderness. Pus may sometimes be seen or elicited at the internal opening. Perianal abscesses are often more readily visible than ischiorectal ones, which can be deeper in the ischiorectal fat and harder to palpate. Digital rectal examination is important when tolerated by the patient. A careful rectal exam can sometimes reveal an internal opening, and care should always be taken to also palpate the side opposite the swelling to gain information as to the possible presence of a bilateral process. Intersphincteric abscesses may be devoid of perianal signs but the patient will be exquisitely tender to digital rectal examination and the diagnosis must be differentiated from a fissure. One additional sign, other than a subtle mass-in-ano and/or pus extrusion, is that an abscess may be associated with inguinal lymphadenopathy whereas a fissure will not.

When the pain is severe but no abscess can be readily detected on physical examination, further

Table 17.2 Anorectal abscess classification

Perianal
Ischiorectal
Intersphincteric
Supralevator

Table 17.3 Parks' classification of fistula-in-ano

Intersphincteric
Transsphincteric
Suprasphincteric
Extrasphincteric

investigations in the form of imaging or examination under anaesthesia are indicated to avoid abscess progression into deeper spaces and/or development of a horseshoe abscess.

Classifications

Anorectal abscesses are classified as listed in Table 17.2. This classification follows from the avenues of infection spread that were discussed in Fig. 17.3. In 1976, Parks and Gordon published the classification of fistula-in-ano listed in Table 17.3 [12].

Special Situation: Fournier's Gangrene

Fournier's gangrene is a rare but potentially devastating condition. It is a mixed microbial infection of aerobic and anaerobic flora, sometimes referred to as a synergistic necrotizing cellulitis, which can have its nidus in the genitourinary tract, gastrointestinal tract or perineal skin. Risk factors include immunocompromise, diabetes, alcohol abuse and paraplegia. The onset can seem like that of a perianal abscess, but the progression is much quicker and far out of keeping with that of a localized abscess. Pain is diffuse, scrotal involvement is common and systemic symptoms and signs can quickly evolve to septic shock and multi-organ failure. Imaging often reveals gas tracking along fascial planes. This diagnosis truly represents a surgical emergency. Management includes a combination of systemic antibiotics and wide surgical debridement, often requiring a second look and sometimes even faecal diversion. The goal of surgical debridement is aggressive resection of skin and underlying tissue back to healthy bleeding tissue. Despite heightened alertness to this condition and expedited care, this condition carries a high mortality rate [13, 14].

Special Situation: The Immunocompromised Host

Anorectal sepsis in the immunocompromised host represents a special circumstance with uncommon presentation. The typical signs and symptoms are predictably less apparent and abscess progression can evolve more quickly. In a recent review of 83 immunocompromised patients with evidence of perianal sepsis, 28 % were HIV positive, 34 % were on steroids for IBD, 20 % had malignancies and 18 % had diabetes. The average age was 44, and 76 % were male. Most patients (40 %) had both an abscess and fistula at the time of presentation. The distribution of fistulas was similar to the general population, although the incidence of horseshoe abscess was higher at 14 %. Healing and complication rates were also comparable to those of the normal population, suggesting that perianal sepsis can be safely managed in immunocompromised patients, provided the diagnosis is made [15]. Patients with neutropenia will often fail to produce a pus-filled abscess and are therefore preferentially treated with antibiotics, unless there is a fluctuant mass mandating incision and drainage in addition to antibiotics [16].

References

1. Goldberg SM, Gordon PH, Nivatvongs S. Essentials of anorectal surgery. Philadelphia: J. B. Lippincott Company; 1980.
2. Morson B, Dawson I. Gastrointestinal pathology. Oxford: Blackwell Scientific Publications; 1972.
3. Hermann G, Desfosses L. Sur la muqueuse de la region cloacale du rectum. C R Seances Acad Sci. 1880;90:1301–4.

4. Parks AG. Pathogenesis and treatment of fistula-in-ano. Br Med J. 1961;1(5224):463–9.
5. Goligher JC, Ellis M, Pissidis AG. A critique of anal glandular infection in the aetiology and treatment of idiopathic anorectal abscesses and fistulas. Br J Surg. 1967;54(12):977–83.
6. Cirocco WC. Life threatening sepsis and mortality following stapled hemorrhoidopexy. Surgery. 2008; 143(6):824–9.
7. van Wensen RJ, van Leuken MH, Bosscha K. Pelvic sepsis after stapled hemorrhoidopexy. World J Gastroenterol. 2008;14(38):5924–6.
8. Hamadani A, Haigh PI, Liu IL, Abbas MA. Who is at risk for developing chronic anal fistula or recurrent anal sepsis after initial perianal abscess? Dis Colon Rectum. 2009;52(2):217–21.
9. Read DR, Abcarian H. A prospective survey of 474 patients with anorectal abscess. Dis Colon Rectum. 1979;22(8):566–8.
10. Ingle SB, Loftus Jr EV. The natural history of perianal Crohn's disease. Dig Liver Dis. 2007;39(10):963–9.
11. Grace RH, Harper IA, Thompson RG. Anorectal sepsis: microbiology in relation to fistula-in-ano. Br J Surg. 1982;69(7):401–3.
12. Parks AG, Gordon PH, Hardcastle JD. A classification of fistula-in-ano. Br J Surg. 1976;63(1):1–12.
13. Rotstein OD, Pruett TL, Simmons RL. Mechanisms of microbial synergy in polymicrobial surgical infections. Rev Infect Dis. 1985;7(2):151–70.
14. Thwaini A, Khan A, Malik A, et al. Fournier's gangrene and its emergency management. Postgrad Med J. 2006;82(970):516–9.
15. Munoz-Villasmil J, Sands L, Hellinger M. Management of perianal sepsis in immunosuppressed patients. Am Surg. 2001;67(5):484–6.
16. North Jr JH, Weber TK, Rodriguez-Bigas MA, Meropol NJ, Petrelli NJ. The management of infectious and noninfectious anorectal complications in patients with leukemia. J Am Coll Surg. 1996; 183(4):322–8.

Imaging of Anal Sepsis

18

Steve Halligan and Stuart A. Taylor

Introduction

Fistual-in-ano describes an abnormal communication between the anal canal (or occasionally the rectum) and the perianal skin. It is a common condition that has a tendency to recur despite apparently curative surgery. Recurrence after surgery is almost always due to infection that has escaped detection by the surgeon and thus gone untreated. It is now increasingly recognised that preoperative imaging, notably by MRI, is able to identify fistulas and associated abscesses that would otherwise have been missed. Not only can MRI elegantly display perianal fistulas, but preoperative MRI has been shown to influence subsequent surgery and significantly diminish the chance of recurrent disease as a result. Because of this, preoperative imaging is becoming increasingly routine, especially in patients with recurrent fistulas.

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Aetiology, Classification and Treatment of Fistula-in-Ano Relevant to Imaging

The previous chapter (Chap. 17) has described the pathophysiology and presentation of anal sepsis. Because cryptogenic fistula-in-ano is predicated by sepsis arising in the intersphincteric plane [1, 2], any useful imaging technique must be able to image this region with precision (i.e. with high spatial and contrast resolution). This is also the case for the patient with an acute perianal abscess, since 87 % may subsequently develop a fistula [3]; imaging during the acute episode (before incision and drainage) may be able to distinguish whether intersphincteric infection underpins the abscess.

By definition a fistula describes an abnormal communication between two epithelial surfaces. The anatomical course of the fistula will be dictated by the location of the infected anal gland and the anatomical planes and structures that surround it. The internal opening of the fistula will usually be in the anal canal at the level of the dentate line, i.e. at the original site of the duct draining the infected gland. In the radial plane, the internal opening is usually posterior at 6 o'clock, simply because anal glands are more abundant posteriorly, especially in men. The dentate line cannot be identified as a discrete anatomical structure by any imaging technique but its position can be approximated with sufficient accuracy by experienced radiologists – it lies approximately 2 cm cranial to the anal verge on coronal images.

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The fistula can reach the perianal skin via a variety of routes, some more tortuous than others and thereby penetrating and involving the muscles of the anal sphincter and surrounding tissues to a variable degree. Fistulas are 'classified' according to the route taken by this 'primary track', which links the internal and external openings. Although there have been a variety of different attempts to classify fistula-in-ano, by far the most widely used is that proposed by Parks and colleagues in 1976 [4]. Parks carefully analysed a consecutive series of 400 patients referred to the surgeons of St. Mark's Hospital London, a specialist hospital dealing with coloproctological disease, and found that he was able to place all fistulas encountered into one of four broad groups: intersphincteric, transsphincteric, suprasphincteric and extrasphincteric [4]. Importantly, most of these groupings could be explained by the cryptoglandular hypothesis. A major role for imaging is the ability to distinguish between these different fistulas and so to arrive at an accurate preoperative classification for the operating surgeon before he or she puts a knife to the skin. Because of this, the interpreting radiologist must be fully conversant with Parks' classification.

While most fistulas probably start as a simple, single primary track, unabated infection may result in ramifications (often multiple) that branch away from this, generally, termed 'extensions'. Extensions are a major target for preoperative imaging because they frequently underpin recurrent disease. This is often because they may occur several centimetres away from the primary track and frequently lie deep in surrounding tissues, thus escaping easy detection. Extensions may be intersphincteric, ischioanal or supralevator (pararectal), and their morphology may suggest tracks or abscesses. Exactly when a 'track' becomes an 'abscess' has no precise definition on imaging.

The ischioanal fossa is the commonest site for an extension, especially one that arises from the apex of a transsphincteric fistula. The ischioanal fossa lies lateral to the sphincter complex, is filled with fat and is traversed by a network of fibroelastic connective tissue. Because this space lies adjacent to the anus (vs the rectum) and lies immediately below (vs above) the levator plate of the pelvic floor, the authors prefer the term 'ischioanal' fossa rather than 'ischiorectal', which is commonly used by surgeons. However, the two terms are interchangeable. Extensions also occur in the horizontal plane and are known as 'horseshoes' if there is ramification of sepsis on both sides of the internal opening.

Although surgical treatment of fistula-in-ano is usually straightforward, most frequently by laying open the fistula, this seemingly simple procedure has many unexpected traps waiting for the unwary. Injudicious incision and overenthusiastic exploration can very quickly convert a simple fistula into a surgical nightmare by creating additional extensions, tracks and communications, with disastrous consequences for the patient. The surgeon's prime objective is to identify the primary track and any associated extensions and then eradicate these by draining all associated infection all while simultaneously preserving anal continence. Thus, there are two surgical questions that should ideally be answered preoperatively:

- What is the relationship between the fistula and the anal sphincter? That is, can the track be safely laid open with only a low risk of post-operative incontinence?
- Are there any extensions from the primary track that need to be treated in order to prevent recurrence? If so, where are they?

Although frequently used for this purpose, it is now well established that examination under (general) anaesthesia (EUA) is not infallible. At EUA, the surgeon attempts to classify the fistula via palpation and probing, so as to determine the relationship to the sphincter. However, the surgeon cannot visualise underlying muscles directly, and general anaesthesia and consequent loss of muscular tone impair precise identification further. The internal opening may be difficult to identify, but probes must not be advanced forcefully for fear of causing unintentional tracks and extensions. For example, forceful probing of a transsphincteric fistula track in the roof of the ischioanal fossa can easily rupture through the levator plate, thereby causing a supralevator extension. In the worst instance, the probe can even rupture into the rectum, converting

a transsphincteric fistula into an extrasphincteric fistula. Although identification of all extensions at EUA is central for cure, missed extensions are the commonest cause of recurrence, reaching 25 % in some series [5].

The net result is that at EUA, it can be very difficult to classify the primary track with confidence, and there is also ample opportunity to make matters worse. Patients with recurrent disease are a particular case in point: They are most likely to harbour foci of missed sepsis but are also most difficult to assess at EUA. In the context of multiple failed operations previously, digital palpation frequently cannot distinguish between scarring due to repeated surgery and induration due to an underlying extension. Furthermore, this group is also most likely to have extensions that travel several centimetres away from the primary track, which further hampers their detection. The more chronic the fistula, the more complicated associated extensions tend to be. The inevitable result is that these patients become progressively more difficult to treat, with both patient and surgeon becoming ever more exasperated. The key to breaking this loop is accurate preoperative imaging.

Imaging of Fistula-in-Ano

For many years radiologists have tried to answer the surgical questions posed in the section above, but with varying degrees of success. Contrast fistulography was the first modality employed. The external opening is catheterised with a fine cannula and water-soluble contrast injected gently in order to define the fistula. Unfortunately, fistulography suffers from two major drawbacks. Firstly, extensions from the primary track may fail to fill with contrast if they are plugged with debris, are very remote, or if there is excessive contrast reflux from either the internal or external openings. Secondly, the sphincter muscles are not imaged directly, which means that the relationship between the fistula and the sphincter must be guessed. Furthermore, inability to visualise the levator plates directly means that it can occasionally be very difficult to decide whether an extension is supra- or infralevator. The net result is that fistulography is both difficult to interpret and its results are unreliable. While initial reports of computerised tomography (CT) for fistula-in-ano were encouraging, simple visualisation of the fistula is not enough; they must be classified correctly, and more mature data suggests that CT cannot do this accurately. This is because the attenuation of the anal sphincter and pelvic floor is similar to the fistula itself unless the latter contains air or contrast, so they cannot be distinguished. This is compounded with a relative inability to image in the surgically relevant coronal plane.

Anal Endosonography

Anal endosonography (AES) was the first technique to directly visualise the anal sphincter complex in detail, and naturally, AES has been applied to the classification of fistula-in-ano (Fig. 18.1). While AES can be very useful, accurate interpretation is highly dependent on the experience of the sonographer. Also, being an ultrasound technique, structures remote from the transducer are difficult to see because penetration of the ultrasound beam is limited. The result is that extensions beyond the

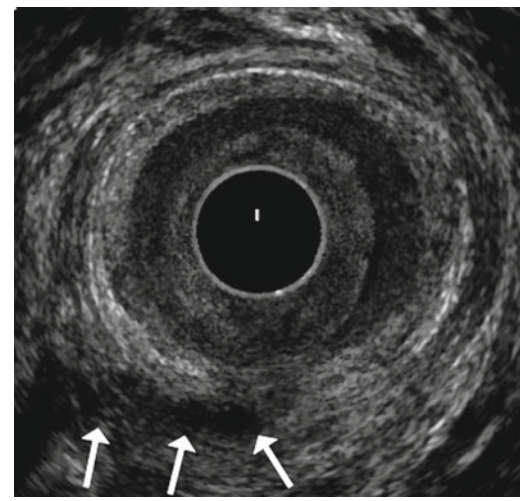

Fig. 18.1 Anal endosonography of a man with a transsphincteric fistula (same patient as Fig. 18.4). There is a hyporeflective track (*arrows*) through the external sphincter, but it is more difficult to appreciate that this is due to a transsphincteric fistula than on the corresponding MR image unless the sonographer is very experienced

sphincter complex are easily missed. Also, AES cannot reliably distinguish infection from fibrosis since both appear hyporeflective, and this causes particular difficulties in patients with recurrent disease since infected tracks and fibrotic scars frequently occur together. While there is no doubt that AES is a valuable technique in the right hands, MRI is generally superior: A study comparing AES to digital evaluation and MRI in 108 primary tracks found that digital evaluation correctly classified 61 %, AES 81 % and MR imaging 90 % [6]. While AES was particularly adept at correctly predicting the site of the internal opening, achieving this in 91 % compared to 97 % for MRI [6], there is little doubt that MRI is a superior technique overall.

MRI Technique

Over the last decade, MRI has emerged as the leading contender for preoperative classification of fistula-in-ano. This is because MRI can vividly separate infected tracks and extensions from surrounding structures, imaging both with precision. Furthermore, MRI can image in the surgically relevant coronal plane so that the geographical course of the fistula can be determined. Indeed, the ability of MRI to not only accurately classify tracks but also to identify disease that would otherwise have been missed has had a palpable effect on surgical treatment and, ultimately, patient outcome [7, 8].

Field strength does not appear to be a critical factor, and excellent results can be obtained using relatively modest MRI scanners with no need for specialised coils. External phased array surface coils increase signal-to-noise ratio (SNR) and spatial resolution, to good effect [9, 10], and are generally available. Although the best spatial resolution is achieved by using dedicated endoluminal anal coils [11], these suffer the same limitation as AES – the limited field of view means that distant extensions will be missed [12]. Because of this, they are now rarely used. It should also be stressed that anal endoluminal coils are not the same as rectal coils. Rather, they are smaller in diameter and are intended to cross the anus.

The MRI sequences used to image fistula-in-ano need to combine anatomic precision (so that the course of the fistula with respect to adjacent structures can be determined), with the facility to highlight sepsis (usually pus). Many investigators employ the rapid and convenient fast spin-echo T2-weighted sequence, which provides good contrast between hyperintense fluid within the track and its hypointense fibrous wall while simultaneously enabling good discrimination between the several layers of the anal sphincter. Fat suppression techniques are very useful. The earliest reports used STIR imaging, with the addition of T1-weighted scans to help anatomical clarification [13], and gadolinium contrast may be used if desired [14]. While other approaches have included saline instillation into the external opening or rectal contrast medium, such measures increase examination complexity in the face of the already excellent results achieved by less invasive procedures, and there is little motivation to adopt them. For the majority of their clinical work, the authors use a 1.5 T magnet and STIR sequences in just two planes, combined with the sagittal acquisition described below, which makes for a very rapid and easy examination.

It is central to success that imaging planes are correctly aligned with respect to the anal sphincter. Because the anal canal is tilted forward from the vertical by approximately 45°, straight axial and coronal images with respect to the patient/scanner tabletop will result in oblique images of the anus, and the geography of any fistula will be difficult to ascertain. This is especially so when trying to determine the height of the internal opening. Oblique axial and coronal planes orientated orthogonal and parallel to the anal sphincter are definitely necessary and are most easily planned from a midline sagittal image (Fig. 18.2). It may be necessary to align supplementary scans to the rectal axis in complex cases with an internal opening high in the rectum, but this is seldom necessary. It is important that the imaged volume extends several centimetres above the levator muscles and also includes the whole presacral space, both of which are common sites for extensions. The entire perineum should also be included. Occasionally, tracks may extend for several centimetres, even leaving the pelvis or reaching the legs, and any track visible on the

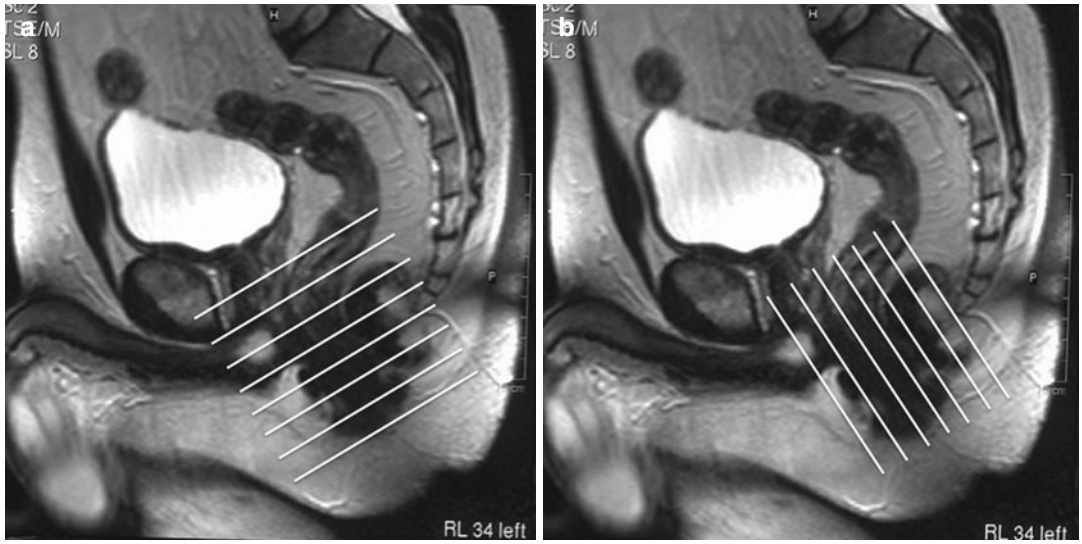

Fig. 18.2 Sagittal T2-weighted planning scan showing the orientation of the anal canal and the oblique axis to which the axial (**a**) and coronal (**b**) scans must be aligned (*white lines*). Images should extend well into the supralevator compartment and also cover the entire presacral space

standard image volume must be followed to its termination if this has not been included. The precise location of the primary track (e.g. ischioanal or intersphincteric) is usually most easily appreciated using axial images, and the radial site of the internal opening is also well seen using this plane. Coronal images best visualise the levator plate, which separates supra- from infralevator infection. The height of the internal opening may also be best appreciated on coronal images, with the caveat that the anal canal must be imaged along its entire craniocaudal extent, as explained above.

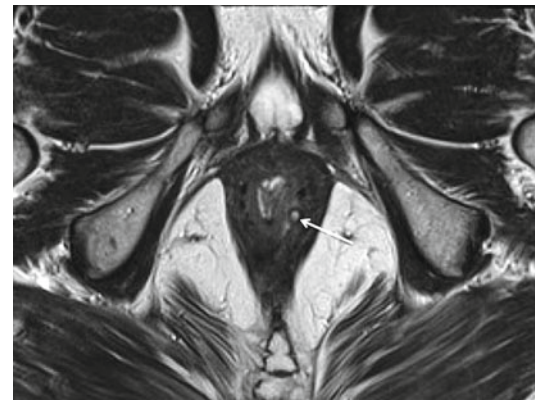

Fig. 18.3 Axial T2-weighted MR images in a man with an intersphincteric fistula (*arrow*). Note that the fistula is contained by the external sphincter; there is no sepsis in the ischioanal fossa

MRI Interpretation

All competent MRI reports should include the following information: The radial location and classification of the primary track(s), the radial location and level of the internal opening(s) and a description of any extensions.

Active tracks are filled with pus and granulation tissue and thus appear as hyperintense on T2-weighted or STIR sequences, often surrounded by hypointense fibrous walls, which can be relatively thick, especially in patients with recurrent disease following previous surgery. The external anal sphincter is relatively hypointense, and its lateral border contrasts sharply against the fat within the ischioanal fossa, especially on T2-weighted studies. Consequently it is relatively easy to determine whether a fistula is contained by the external sphincter or has extended beyond it.

If a fistula remains contained by the external sphincter throughout its course, then it is highly likely to be intersphincteric (Fig. 18.3).

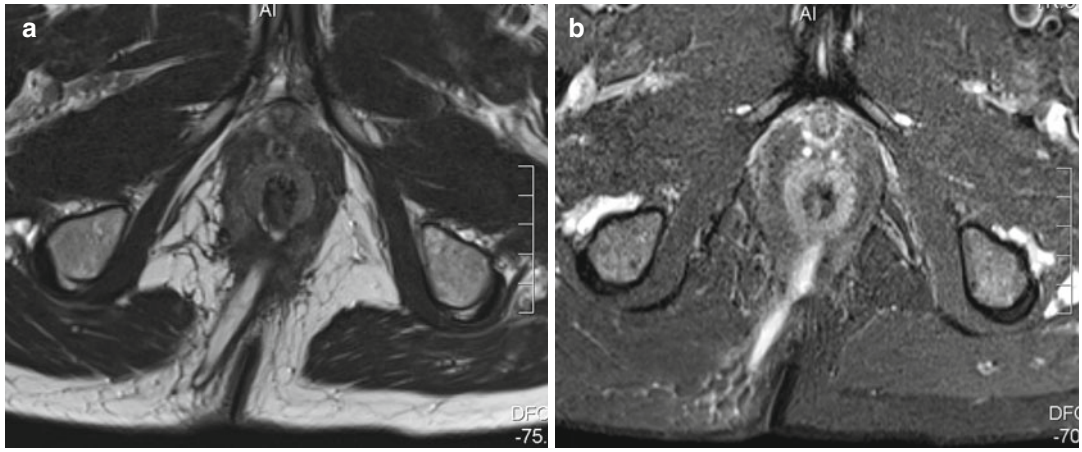

Fig. 18.4 Axial T2-weighted (**a**) and STIR (**b**) images in a man with a transsphincteric fistula in the right posterior quadrant with an internal opening at 6 o'clock at dentate line level (same patient as Fig. 18.1)

In contrast, transsphincteric, suprasphincteric and extrasphincteric fistulas all show sepsis in the ischioanal space – it is the level of the internal opening and the level at which the fistula crosses the sphincter complex that differentiates between these types. A track in the ischioanal fossa is usually due to a transsphincteric fistula (Fig. 18.4) simply because it is much commoner than suprasphincteric or extrasphincteric classifications.

The exact location of the internal opening can be difficult to define. Two questions need to be answered: what is the radial site of the internal opening and what is its level? The vast majority of anal fistulas open into the anal canal at the level of the dentate line, commensurate with the cryptoglandular hypothesis of fistula pathogenesis. Furthermore, most fistulas also enter posteriorly, at 6 o'clock. Although the dentate line cannot be identified as a discrete anatomical entity, even when using endoanal receiver coils, its general position can be estimated with sufficient precision by an experienced radiologist. The dentate line lies at approximately mid-anal canal level, which is generally midway between the superior border of the puborectalis muscle and the most caudal extent of the subcutaneous external sphincter. These landmarks define the 'surgical' anal canal (as distinct from the 'anatomical' anal canal, which is shorter, and defined as the canal caudal to the anal valves). Dentate level is best estimated using coronal views, which allow the craniocaudal extent of the puborectalis muscle and external sphincter to be appreciated, but its location can also be estimated from axial views given sufficient experience. It should be noted that in many patients the puborectalis muscle is rather gracile, unlike the bulky muscle suggested in many anatomical illustrations. Notably, the puborectalis frequently blends imperceptibly into the external sphincter, which hampers precise identification of mid-anal canal level on imaging. Nevertheless, this can be overcome with experience. Any fistula track that penetrates the pelvic floor above the level of the puborectalis muscle is potentially a suprasphincteric or extrasphincteric fistula. The level of the internal opening distinguishes between these, being anal in the former (Fig. 18.5) and rectal in the latter (Fig. 18.6). Transsphincteric fistulas penetrate the external sphincter directly, a feature that can be easily appreciated on axial or coronal views (Fig. 18.4). However, recent MR studies have revealed that a transsphincteric track may cross the sphincter at a variety of angles [15]. For example, it may arch upwards as it passes through the external sphincter and thus cross the muscle at a higher level than would be deduced merely from inspecting the level of the internal opening. This is important because such tracks will require a greater degree of sphincter incision during fistulotomy, with a correspondingly increased risk of post-operative incontinence. Coronal MRI is best placed to estimate the

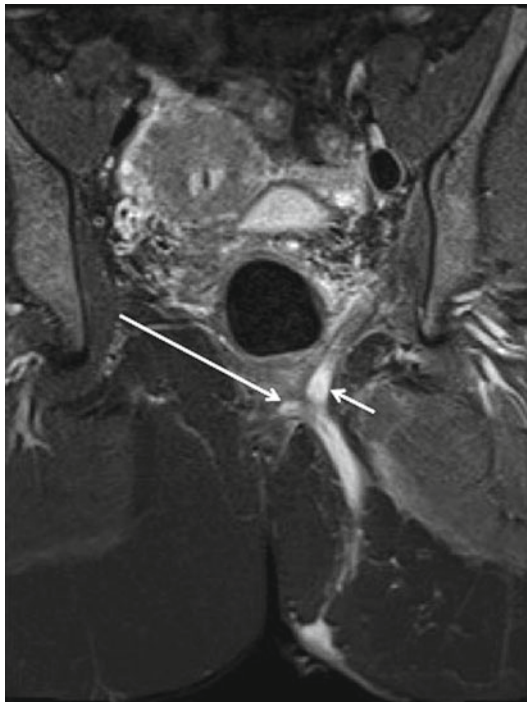

Fig. 18.5 Coronal STIR MRI in a patient with a suprasphincteric fistula. The primary track is arching over the puborectalis (*long arrow*). There is also a small cranial extension off the apex of the track (*short arrow*) into the roof of the left ischioanal fossa

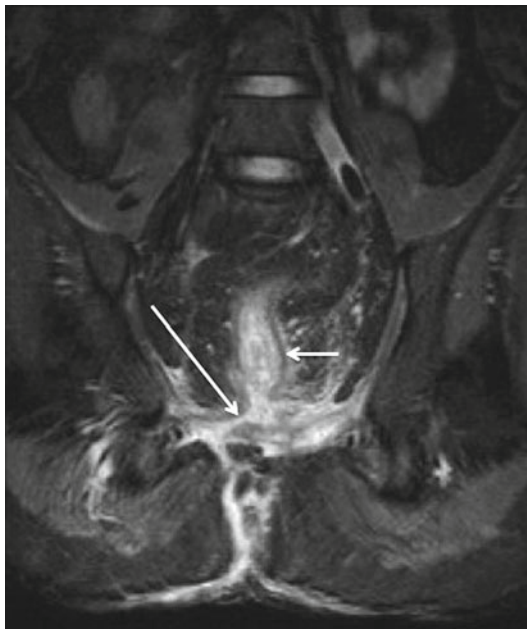

Fig. 18.6 Coronal STIR MRI in a man with an extrasphincteric fistula. Note that the primary fistula track has a rectal opening (*long arrow*) above the level of the levator plates (rectum = *short arrow*)

precise angulation of the track with respect to the surrounding musculature [15].

The radial site of the internal opening is simple to identify if the fistula track can be traced right to the anal mucosa, but this is unusual because the internal opening is rarely widely patent – rather it is most often compressed and can be very difficult to see. In many cases an intelligent deduction must be made as to where the internal opening is likely to be. This is best achieved by looking to where there is maximal intersphincteric sepsis, since the internal opening is likely to lie adjacent or very close to this. The intersphincteric space and longitudinal layer are often seen as a low intensity ring lying between the internal and external sphincter. The internal sphincter is hyperintense on both T2-weighted fast spin echo and STIR sequences.

Extensions

The major advantage of MRI is the ease with which it can image any extensions associated with the primary track. Like tracks, extensions are manifest as hyperintense regions on T2-weighted and STIR imaging and also enhance further if intravenous contrast is given. Again, collateral inflammation can be present to variable extent. The commonest type of extension is one that arises from the apex of a transsphincteric track and extends into the roof of the ischioanal fossa (Fig. 18.7).

The major benefit of preoperative MRI is that it can alert the surgeon to extensions that would otherwise be missed during EUA. This is especially the case when extensions are either contralateral to the primary track or when they are several centimetres away from it (Fig. 18.8). It is especially important to image supralevator extensions (Fig. 18.9) since these are not only particularly difficult for the surgeon to detect, but they also pose specific difficulties with treatment. Horseshoe extensions spread to either side of the internal opening and are recognised on MRI by their unique configuration (Fig. 18.10). Horseshoes may be intersphincteric, ischioanal or supralevator. Complex extensions are especially common in patients with recurrent fistula-in-ano or those who have Crohn's disease.

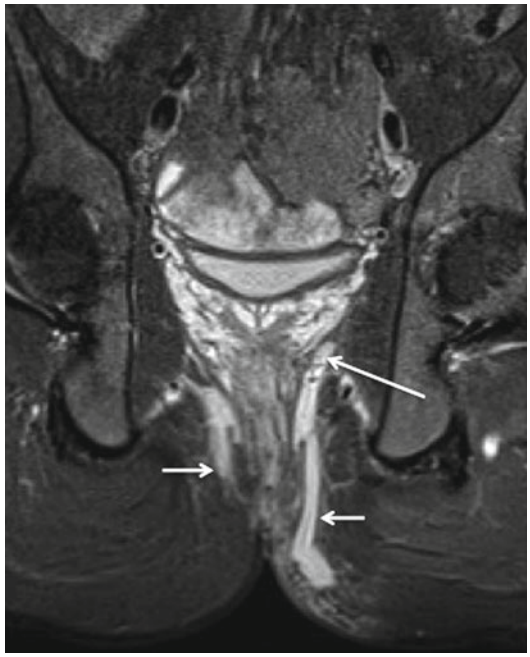

Fig. 18.7 Coronal STIR MRI in a patient with bilateral transsphincteric fistulas (*short arrows*). There is an extension (*long arrow*) from the apex of the left fistula into the roof of the left ischioanal fossa

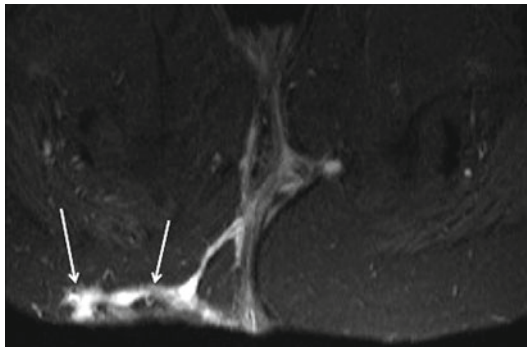

Fig. 18.8 Axial STIR MRI in a patient with extensions (*arrows*) into the right buttock, several cm from the anus

Effect of Preoperative MRI on Surgery and Clinical Outcome

Over the last decade, imaging, notably MRI, has revolutionised the treatment of patients with fistula-in-ano. As stated in the sections above, this is because MRI can preoperatively classify fistulas with high accuracy while also alerting the surgeon to disease that would otherwise have been missed. While there are reports of the technique dating from 1989 [16], it was not until the description by Lunniss and co-workers [13] that the true potential of MRI was appreciated fully. Lunniss imaged 16 patients with cryptoglandular fistula-in-ano and compared the classification achieved by MRI with that obtained at subsequent EUA. MRI proved correct in 14 of the 16 cases (88 %), immediately suggesting that it was by far the most accurate preoperative assessment yet available. However, the remaining two patients, in whom MRI had suggested disease in

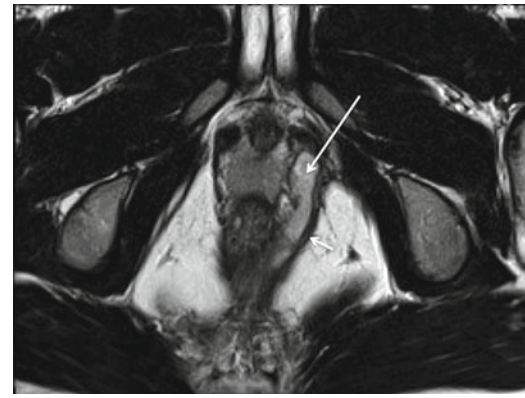

Fig. 18.9 Axial T2-weighted MRI in a patient with a left-sided supralevator extension (*long arrow*). Note the extension is supralevator because it lies medial to the left levator plate (*short arrow*)

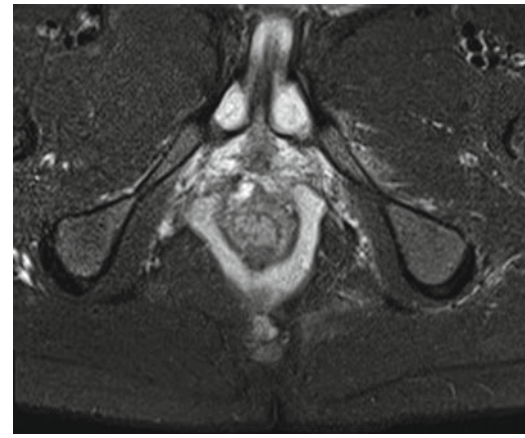

Fig. 18.10 Axial STIR MRI in a patient with a typical horseshoe extension wrapping around both posterior quadrants of the anus

the face of a normal EUA, represented some months later with disease at the site initially indicated by MRI. The clear implication was that EUA had missed disease that had been detected by MRI. This led the authors to conclude, 'MRI is the most accurate method for determining the presence and course of anal fistulae' [13].

Lunniss' work was rapidly confirmed by others working in the field and subsequently elaborated on. For example, Spencer and colleagues independently classified 37 patients into those with simple or complex fistulas on the basis of MRI and EUA and found that imaging was the better predictor of outcome, with positive and negative predictive values of 73 % versus 57 % and 87 % versus 64 % for MRI and surgery, respectively [17]. This study implied clearly that MRI and clinical outcome were closely related and again raised the possibility that preoperative MRI could help identify features that caused post-operative recurrence. Beets-Tan and colleagues extended this hypothesis by investigating the therapeutic impact of preoperative MRI; the MRI findings in 56 patients were revealed to the operating surgeon after they had completed an initial EUA [9]. MRI provided important additional information that precipitated further surgery in 12 of the 56 patients (21 %), mostly in those with recurrent fistulas or Crohn's disease [9]. Buchanan and co-workers hypothesised that the therapeutic impact and thus beneficial effect of preoperative MRI would be greatest in patients with recurrent fistulas, since these had the most chance of harbouring occult infection while simultaneously being the most difficult to evaluate clinically [10]. After an initial EUA, they revealed the findings of preoperative MRI in 71 patients with recurrent fistulas and left any further surgery performed in the light of the MRI findings to the discretion of the operating surgeon. The clinical course of each patient was then followed subsequently. They found that post-operative recurrence was only 16 % for surgeons who always acted when MRI suggested they had missed areas of sepsis, whereas recurrence was 57 % for those surgeons who always chose to ignore imaging, believing their own assessment to be superior [10]. Furthermore, of the 16 patients who needed further unplanned surgery, MRI initially correctly predicted the site of this disease in all cases [10].

Ever since Lunniss' work suggested that EUA might be an imperfect reference standard with which to judge MRI [13], comparative studies have been plagued by the lack of a genuine reference standard. It is now well recognised that surgical findings at EUA are sometimes incorrect. In particular, false-negative diagnoses are relatively frequent. In a recent comparative study of endosonography, MRI and EUA in 34 patients with fistulas due to Crohn's disease, Schwartz and co-workers found that a combination of the results of at least two modalities was necessary in order to arrive at a correct classification [18]. Because surgical false-negatives will only reveal themselves over the course of long-term clinical follow-up, comparative studies that ignore clinical outcome are likely to be seriously flawed. Recognising this, Buchanan and co-workers examined 108 primary tracks by digital examination, anal endosonography and MRI and then followed patients' clinical progress to establish an enhanced reference standard for each patient that was based on ultimate clinical outcome rather than EUA [6]. The authors found that digital evaluation correctly classified 61 % of primary tracks, AES 81 % and MR imaging 90 % [6]. While endosonography was particularly adept at predicting the site of the internal opening correctly, achieving this in 91 %, MRI was even better at 97 % and was superior to endosonography in all assessments investigated by the authors [6]. While endosonography is certainly a useful tool for investigating fistula-in-ano, it cannot compete with MRI for detection of extensions, which is undoubtedly the most important role for preoperative imaging. MRI is also more generally available and less operator dependent.

Differential Diagnosis of Perianal Sepsis

Not all perianal sepsis is caused by fistula-in-ano. For example, acne conglobata, hidradenitis suppurativa, pilonidal sinus, actinomycosis,

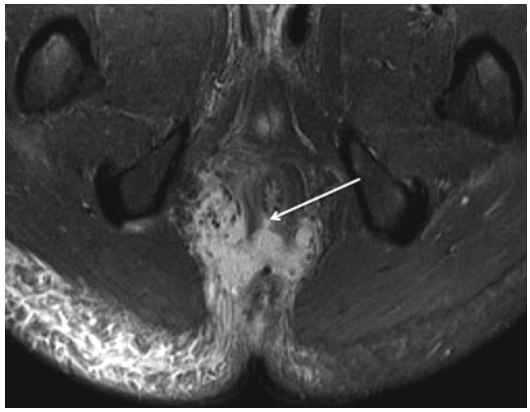

Fig. 18.11 Axial STIR image in a patient with an acute perianal abscess. Note that there is a communication with the intersphincteric plane posteriorly at 6 o'clock (*arrow*), suggesting that the abscess is due to cryptoglandular infection and that the patient may subsequently develop a fistula

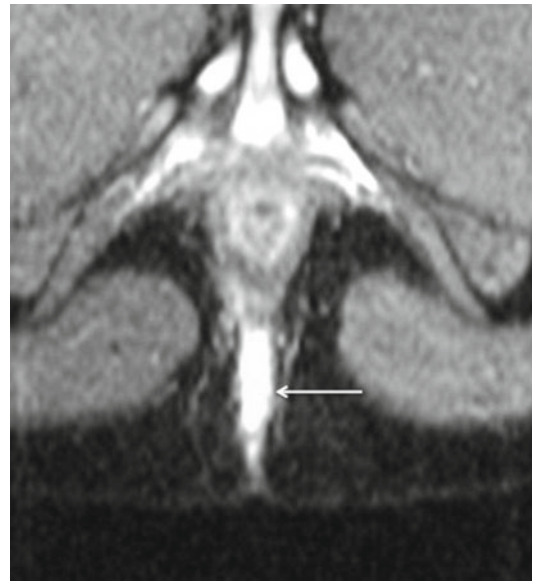

Fig. 18.12 Axial STIR MRI in a patient with pilonidal sinus. There is posterior sepsis (*arrow*) but this stops at the posterior margin of the external sphincter and does not enter the intersphincteric plane

tuberculosis, proctitis, human immunodeficiency virus, lymphoma and anal and rectal carcinoma may all cause perianal infection. While clinical examination is often conclusive, this is not always the case and imaging may help with differential diagnosis. The cardinal feature of fistula-in-ano is intersphincteric infection, which is not generally found in other conditions, although it may be detected if MRI is used to image acute anorectal abscesses (Fig. 18.11). Whenever imaging suggests that infection is superficial rather than deep seated and that there is no sphincteric involvement, then other conditions such as hidradenitis suppurativa should be considered. For example, a study comparing patients with pilonidal sinus (Fig. 18.12) and fistula-in-ano found that MRI could reliably distinguish between the two on the basis of intersphincteric infection and an enteric opening, both of which were always absent in pilonidal sinus [19].

The possibility of underlying Crohn's disease should always be considered in patients who have particularly complex fistulas, especially if the history is relatively short. Indeed, a perianal fistula is the presenting symptom in approximately 5 % of patients, and 30–40 % will experience anal disease at some time [18, 20]. The MRI examination can be extended cranially to encompass the small bowel where Crohn's disease is suspected, and the possibility of underlying pelvic disease should be considered in any patient with an extrasphincteric fistula, whether thought due to Crohn's disease or otherwise.

Which Patients Should Be Imaged?

While most patients with fistula-in-ano are simple to both diagnose and treat, a proportion will benefit from detailed and accurate preoperative investigation. Where there is easy access to MRI, it could be argued that all patients should be imaged preoperatively. For example, while the therapeutic impact of preoperative MRI is undoubted in patients with complex disease [9, 10], it has been estimated that the therapeutic impact of MR imaging is 10 % in patients presenting for the first time with seemingly simple fistulas [21]. However, where access to imaging is more restricted, the clinician and radiologist will need to select those patients who are most likely to benefit. Since there is now overwhelming evidence that MRI alters surgical therapy and improves clinical outcome in patients with

recurrent disease, MRI should be routine in such cases. Patients presenting for the first time with a fistula that appears complex on clinical examination should also be referred, as should patients with known Crohn's disease since the preponderance of complex fistulas is increased in this situation.

There are also surgical situations where imaging is likely to be particularly beneficial, even when the fistula itself is simple. For example, the anterior external sphincter is very short in women and dividing this during fistulotomy particularly risks post-operative incontinence, even when the fistula itself is simple and has no extensions. Faced with such a dilemma, rather than incising the fistula, the surgeon may choose to pass a seton through the track in order to provide drainage. The patient can then be imaged post-operatively in order to assess the potential extent of sphincter division by visualising the relationship of the seton to the external sphincter. A decision can then be made whether to progress with fistulotomy or to keep the seton in place for a few months, after which time the internal opening can be closed with a rectal mucosal advancement flap. Setons may also be placed at EUA when the surgeon is uncertain about the relationship between the track and the sphincter and then imaged post-operatively in order to answer this question if imaging has not been performed preoperatively.

The benefit of MRI is not restricted to surgical assessment. The advent of monoclonal antibody to human tumour necrosis factor alpha has impacted dramatically upon the medical management of patients whose fistulas are due to Crohn's disease, especially those with chronic disease. However, therapy is contraindicated if an abscess is present and MRI may be used to search for this. Indeed, MRI may be used to monitor therapy since it seems that fistulas may persist in the face of clinical findings that suggest remission. For example, MRI studies in patients whose external opening has closed have revealed that underlying sepsis is often still present, indicating a need for continuing therapy [22].

Conclusion

In those patients with fistula-in-ano who have a high likelihood of complex disease, the evidence that preoperative MRI influences the surgical approach and extent of exploration and improves ultimate outcome is now overwhelming. We hope that this chapter will stimulate both surgeons to ask for this service and for radiologists to provide it. Doing so will reduce the incidence of recurrent fistula-in-ano and the misery this causes.

References

1. Chiari H. Uber die analen divertikel der rectumschleimhaut und ihre beziehung zu den anal fisteln. Wien Med Press. 1878;19:1482–3.
2. Parks AG. The pathogenesis and treatment of fistula-in-ano. Br Med J. 1961;5224:463–9.
3. Fucini C. One stage treatment of anal abscesses and fistulae. Int J Colorectal Dis. 1991;6:12–6.
4. Parks AG, Gordon PH, Hardcastle JD. A classification of fistula-in-ano. Br J Surg. 1976;63:1–12.
5. Sanio P. Fistula-in-ano in a defined population: incidence and epidemiological aspects. Acta Chir Gynaecol. 1984;73:219–24.
6. Buchanan G, Halligan S, Bartram CI, Williams AB, Tarroni D, Cohen CRG. Clinical examination, endosonography, and magnetic resonance imaging for preoperative assessment of fistula-in-ano: comparison to an outcome derived reference standard. Radiology. 2004;233:674–81.
7. Halligan S, Buchanan G. Imaging fistula-in-ano. Eur J Radiol. 2003;42:98–107.
8. Halligan S, Stoker J. Imaging fistula-in-ano: state-of-the-art. Radiology. 2006;239:18–33.
9. Beets-Tan RG, Beets GL, van der Hoop AG, Kessels AG, Vliegen RF, Baeten CG, van Engelshoven JM. Preoperative MR imaging of anal fistulas: does it really help the surgeon? Radiology. 2001;218:75–84.
10. Buchanan G, Halligan S, Williams A, Cohen CR, Tarroni D, Phillips RK, Bartram CI. Effect of MRI on clinical outcome of recurrent fistula-in-ano. Lancet. 2002;360:1661–2.
11. Hussain SM, Stoker J, Schouten WR, Hop WC, Lameris JS. Fistula in ano: endoanal sonography versus endoanal MR imaging in classification. Radiology. 1996;200:475–81.
12. Halligan S, Bartram CI. MR imaging of fistula in ano: are endoanal coils the gold standard? AJR Am J Roentgenol. 1998;171:407–12.
13. Lunniss PJ, Armstrong P, Barker PG, et al. Magnetic resonance imaging of anal fistulae. Lancet. 1992;340:394–6.

14. Spencer JA, Ward J, Beckingham IJ, Adams C, Ambrose NS. Dynamic contrast-enhanced MR imaging of perianal fistulas. Am J Roentgenol. 1996;167:735–41.
15. Buchanan GN, Williams AB, Bartram CI, Halligan S, Nicholls RJ, Cohen CRG. Potential clinical implications of direction of a trans-sphincteric anal fistula track. Br J Surg. 2003;90:1250–5.
16. Koelbel G, Schmiedl U, Majer MC, et al. Diagnosis of fistulae and sinus tracts in patients with Crohn disease: value of MR imaging. Am J Roentgenol. 1989; 152:999–1003.
17. Spencer JA, Chapple K, Wilson D, Ward J, Windsor AC, Ambrose NS. Outcome after surgery for perianal fistula: predictive value of MR imaging. Am J Roentgenol. 1998;171:403–6.
18. Schwartz DA, Wiersema MJ, Dudiak KM, et al. A comparison of endoscopic ultrasound, magnetic resonance imaging, and exam under anesthesia for evaluation of Crohn's perianal fistulas. Gastroenterology. 2001;121:1064–72.
19. Taylor SA, Halligan S, Bartram CI. Pilonidal sinus disease: MR imaging distinction from fistula in ano. Radiology. 2003;226:662–7.
20. Schwartz DA, Loftus Jr EV, Tremaine WJ, et al. The natural history of fistulizing Crohn's disease in Olmsted County, Minnesota. Gastroenterology. 2002; 122:875–80.
21. Buchanan GN, Halligan S, Williams AB, Cohen RCG, Tarroni D, Phillips RKS, Bartram CI. Magnetic resonance imaging for primary fistula in ano. Br J Surg. 2003;90:877–81.
22. Bell SJ, Halligan S, Windsor ACJ, Williams AB, Wiesel P, Kamm A. Response of fistulating Crohn's disease to infliximab treatment assessed by magnetic resonance imaging. Aliment Pharmacol Ther. 2003; 17:387–93.

Management of Anorectal Sepsis

19

Toby M. Hammond and Peter J. Lunniss

Introduction

Anorectal sepsis is common, presenting as either an acute abscess or a chronically discharging fistula. The majority can be managed without complication, but a proportion of those with chronic sepsis can present a major challenge to both patient and surgeon. This was recognised by Hippocrates in 460 BC and was further emphasised by reports from the middle ages of clinicians whose primary function was to treat anal fistulas and the highest surgical fee in history being paid for the successful treatment of the anal fistula of Louis XIV.

In the UK, the majority (>90 %) of those with chronic anorectal sepsis have no proven underlying aetiology, and the condition is classified as non-specific, idiopathic or cryptoglandular, the diseased anal gland in the intersphincteric space deemed central to its evolution. The remainder of cases may be seen in association with inflammatory bowel disease (predominantly Crohn's disease), tuberculosis, pilonidal disease, hidradenitis suppurativa, malignancy, trauma (accidental or iatrogenic) and foreign bodies [1]. Although most chronic anal fistulas are preceded by an acute abscess, around 40 % of acute abscesses are caused by suppurative skin conditions, almost invariably of staphylococcal origin, and which have nothing to do with fistulae.

Anorectal sepsis most commonly afflicts people in their third to fifth decade, with a male predominance (2:1–4:1). Anorectal abscesses are among the commonest surgical emergencies and require prompt operative drainage. In England and Scotland, the annual incidence of such procedures is approximately 1 in 5,000 of the population [2]. The most accurate data on the incidence of idiopathic anal fistulas is from Scandinavia, which shows an annual incidence of 8.6–10/100,000 [3].

Acute Anorectal Sepsis

The approach to the management of acute anorectal sepsis should be twofold:

1. Determine and define the underlying pathology.
2. Ensure all septic collections are drained, and if sufficiently experienced, identify any fistulous tracts and consider insertion of a loose seton or primary fistulotomy.

Aetiology can usually be determined by the history and clinical examination, examination under anaesthesia including rigid sigmoidoscopy and microbiological and histological

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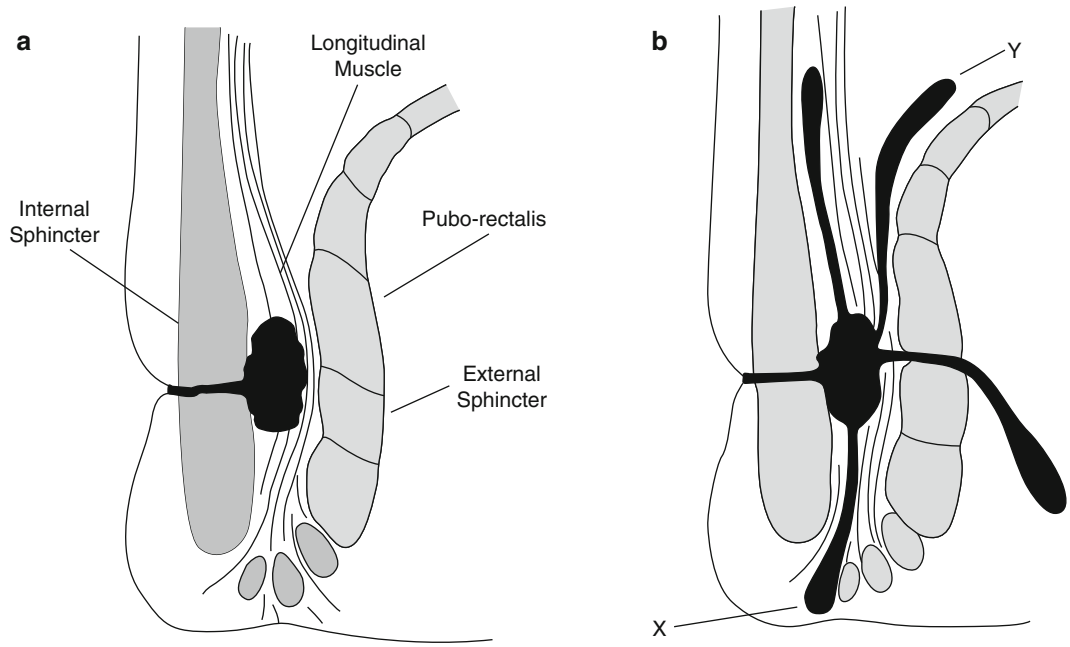

Fig. 19.1 Illustration of the spread of infection from the primary anal gland abscess (**a**) into the surrounding tissues. (**b**) The commonest course is that marked X and the most rare Y (Reproduced from Parks [4], with permission from BMJ Publishing Group Ltd)

appraisal. Pus in the superficial perianal space may result from a simple cutaneous infection or the caudal spread of an intersphincteric abscess. Pus spreads along anatomical planes of least resistance, provided by the branching extensions of the conjoined longitudinal muscle layer. Cephalad extension of an intersphincteric abscess will result in a high intermuscular or supralevator abscess. An abscess in the ischiorectal space may result from a non-cryptoglandular cause, or the lateral spread of sepsis from intersphincteric sepsis having crossed the external sphincter (Fig. 19.1). Upward spread will traverse the levators to cause a supralevator abscess [4]. Circumferential spread can occur in the intersphincteric, ischiorectal or supralevator (pararectal) planes resulting in a horseshoe extension [5].

Traditionally, acute anorectal sepsis is treated by incision and drainage, there being no convincing evidence to recommend a simple incision over skin excision, in the form of saucerisation or an ellipse. The authors' preference however is for an elliptical excision of affected skin, it being intuitive that simple incision alone may allow for early wound closure and therefore premature recurrence of sepsis, whilst saucerisation creates a larger than necessary wound which may result in prolonged healing with poorer cosmesis. The wound is lightly tucked, ensuring the dressing reaches the wound apices.

Microbiology allows a logical staged approach to treatment. At drainage, pus should be sent for culture. The presence of skin-derived organisms alone effectively rules out a cryptoglandular aetiology and therefore recurrence of sepsis and fistula. Culture of bowel-related organisms suggests an underlying fistula, and such patients should be followed up and informed of the likelihood of recurrent sepsis. Although 100 % sensitive for the presence of a fistula, microbiological results cannot predict whether a fistula will chronically persist, in contrast to the finding of intersphincteric space sepsis at the time of drainage [6].

The results of microbiological culture are obviously not available at the time of initial surgery, and the patient may not always be best served by incision and drainage alone. The

cryptoglandular hypothesis holds eradication of the diseased intersphincteric anal gland as a sine qua non of success and if not adequately treated in the early stages can lead to a more complex fistula [7]. Primary fistulotomy, in the presence of intersphincteric space sepsis, therefore has a number of proponents but carries risk. Inflammation and oedema may obscure the internal opening, resulting in only one-third being evident at the time of initial surgery, and in conjunction with overzealous probing, can lead to the creation of false tracks. Fistulotomy can cause flatus incontinence and passive soiling, which may be unacceptable to a number of patients, some of whom may never have developed subsequent recurrent sepsis. A Cochrane review concluded that primary fistulotomy with abscess drainage significantly reduced recurrence or persistence of the fistulous abscess, and the need for repeat surgery, and that there was no statistically significant evidence of incontinence following fistulotomy [8]. The authors would nonetheless suggest a judicious approach to such a policy and recommend simple incision and drainage when a fistula is not evident and primary fistulotomy in those with a clearly delineated low fistula if the patient is agreeable to such a strategy. If there are concerns regarding continence, a loosely tied seton will allow a more considered approach at a later time.

Chronic Anal Fistulas

Management of the chronic anal fistula is necessarily more diverse than its acute predecessor and depends upon accurate anatomical knowledge of the fistula's course through the anal sphincter complex. Failure to appreciate the importance of this relationship may result in fistula recurrence, incontinence or, catastrophically, both. Classification of the pathology is of the upmost importance, as it will guide surgical management.

The most comprehensive, practical and widely used classification is that devised by the late Sir Alan Parks [5]. It is based on the cryptoglandular hypothesis and the relationship of the primary fistula track to the external sphincter. Four main groups exist: intersphincteric, transsphincteric, suprasphincteric and extrasphincteric. These groups can be further subdivided according to the presence and course of any extensions or secondary tracks.

Intersphincteric fistulas (55 %) are usually simple tracks (uncomplicated fistulas consisting only of the primary track) passing down through the intersphincteric space to the perianal skin; but others may have a high blind track, a secondary high opening into the rectum or no perianal opening or even a high pelvic extension.

Transsphincteric fistulas (40 %) cross the external sphincter to pass through the ischiorectal fossa to reach the gluteal skin. They may be subdivided into 'high', 'mid' or 'low' dependant on where the track crosses: above, at the level of or below the dentate line, respectively. This may not be at the same level the track crosses the internal sphincter. Fistulas may be simple or have a high blind track terminating above or below the levator ani muscles.

Suprasphincteric fistulas (<5 %) run up the intersphincteric space to a level beyond the puborectalis and then curl over it through the levator ani and into the ischiorectal fossa to reach the skin. An argument exists as to whether suprasphincteric tracks can be part of this classification, as some believe that they are in fact very high transsphincteric tracks or have been created iatrogenically.

Extrasphincteric fistulas are not of cryptoglandular pathology. They run without relation to the sphincters and are classified according to their pathogenesis.

Clinical Assessment

A full history and examination, including proctosigmoidoscopy, are essential in all cases to assist in determining the aetiology of the fistula. Clinical assessment involves five essential points: location of the internal opening, location of the external opening, the course of the primary track, the presence of any secondary extensions and the presence of other diseases complicating the fistula.

Digital assessment of the primary track, by an experienced coloproctologist in the conscious patient, has been shown to be 85 % accurate [9]. It is further complimented by examination under anaesthesia (EUA), involving the same careful digital examination, followed (before any judicious probing) by the instillation of dilute hydrogen peroxide via the external opening to identify the internal opening.

Although careful examination under anaesthetic is the most important part of any assessment, previous surgery can lead to scarring and deformity, and complex fistulas with multiple secondary tracks make clinical assessment difficult. In such situations imaging is indicated, and in the case of fistula recurrence or concern regarding the patient's continence, baseline assessment of sphincter integrity and strength is recommended. Surgery can then be tailored to the individual, and the patient appropriately consented – one patient's minor soiling may be the cause of another's social isolation.

Treatment Options

The management of anal fistulas has traditionally been purely surgical. The wide range of surgical techniques developed is testament to the fact that none are universally effective at achieving the dual aims of permanent fistula eradication and the preservation of sphincter function. At one end of the spectrum, fistula eradication is best achieved by fistulotomy, and at the other, no attempt at complete eradication of the fistula, but rather palliation of symptoms, using a loose drainage seton. Various strategies have been adopted that lie between these two extremes, which can be divided between those that still divide the sphincters, but attempt to minimise the functional consequences, including the therapeutic use of setons, and those which attempt to preserve sphincteric function. These include the use of advancement flaps, procedures that target the intersphincteric component of the fistula tract (the 'LIFT' procedure) and biomaterials that can act as a scaffold for tissue repair. Unfortunately, it is virtually impossible from the literature to determine any meaningful comparisons between strategies as patient demographics (including previous obstetric history, fistula or other anal surgery) are either inadequately reported or vary considerably, results are often not reported relative to the specific type of fistula aetiology or classification, and interpretation of fistula classification may vary (one surgeon's 'high fistula' is another's 'low fistula'). Additionally reports of success tend not to be equalled by reporting of failures or functional disturbance; and most reports have inadequate follow-up. Furthermore, in such a field of surgery, the use of prospective randomised controlled trials is problematic due to the heterogeneity of fistula anatomy, individual surgeon treatment preference and the ethical difficulties of comparing treatment strategies in which functional outcomes may be markedly different. Levels of evidence are thus generally poor (the potential influence of industry in a specialty which should be industry assisted is beyond the remit of this chapter).

Nonetheless, it is well recognised that critical to the success of all techniques are the elimination of acute sepsis and the eradication of any secondary extensions [10]. On theoretical grounds, attention to the primary tract is also necessary to optimise the chances of success. The aim should be to convert the chronically inflamed or epithelialised tract, often surrounded by dense fibrosis, to an acute wound, thereby allowing the healing cascade to recommence with the potential for progression to complete tissue repair. Failure to adequately remove all granulation or epithelial tissue will affect fibroblast and endothelial cell migration and, possibly in conjunction with inadequate removal of the presumed source (the diseased intersphincteric anal gland), will inhibit healing [11].

Sphincter Dividing Techniques

Fistulotomy

This technique, dating back at least to the fourteenth century, involves surgically dividing the tissue enclosed by the fistula tract and allowing the wound to heal by secondary intention. To date

it is the most effective method of fistula eradication but through division of the enclosed sphincter muscle fibres renders the patient at risk of continence disturbance, with reported rates ranging from 5 to 40 % [12]. Fistulotomy is thus usually reserved for those patients in whom the consequences of sphincter division are anticipated to result in no or minimal functional disturbance, for example, 'low' fistulas, traditionally interpreted as intersphincteric and transsphincteric tracks, the latter involving <30 % of the external sphincter, but not anteriorly in women. Additionally, it is suitable for those patients, preferably with good preoperative function and strong anal sphincters, who are prepared to risk continence to be rid of their symptoms.

Marsupialisation (suturing the divided wound edges to the edges of the fistula track) and fistulotomy followed by immediate reconstruction of the divided muscle have been described in attempts to improve wound healing and continence disturbance, respectively. They have both achieved good results in comparison to conventional fistulotomy.

Staged Fistulotomy

An alternative treatment for 'higher' fistulas is a two-stage fistulotomy, in which, initially, part of the sphincter beneath the primary track is divided and a loose seton placed across the remaining sphincter. This aims to reduce the consequences of sphincter division at a single stage, by allowing fibrosis in the area of division and therefore theoretically reducing retraction of that muscle divided at the second stage. This technique is associated with high levels of fistula eradication rates but is no longer popular as the functional consequences (when reported) are highly variable.

The loose seton has been used with the aim of entire external sphincter preservation in transsphincteric fistulas. The internal sphincter is divided up to the level of the internal opening (thereby removing the infecting source, the chronically inflamed anal gland in the intersphincteric space), and the external sphincter is enclosed within a loose seton. This is removed once all the wounds have healed satisfactorily (this occurs in about 60 % of patients), to allow spontaneous healing of the remaining tract. However, this technique has two drawbacks, the functional consequences of internal sphincter division and high rates of fistula recurrence in the long term [13].

The Tight Seton

The tight or cutting seton (derived from the Latin word 'seta', meaning a bristle) has been used in the surgical management of anal fistulas since the time of Hippocrates, who recorded the use of a horse hair thread, tightened intermittently 'until the enclosed flesh was eaten through'. The rationale of the technique is similar to that of the staged fistulotomy, in that the sphincter complex is gradually severed, by repeated tightening or replacement of the seton, followed by fibrosis which supposedly prevents the divided muscle springing apart.

Direct comparisons of the published data are difficult due to the heterogeneity of fistula aetiology and anatomy, whether the first stage incorporates internal sphincterotomy, the seton material employed (which includes stainless steel, rubber bands, a variety of braided and monofilament sutures and plastic cable ties), interval and frequency of tightening, the time taken for the seton to cut through the encircled tissue and the varying lengths of follow-up. However, irrespective of the material used, the technique is nearly always associated with successful eradication of the fistula, but also frequently at the expense of anal continence. The rates of incontinence associated with the conventional cutting seton are likely to be proportional to the speed of sphincter division, in that the tighter the seton, the quicker the enclosed tissue is divided which leaves less time for fibrosis to develop and therefore the greater the sphincter disruption. Additionally, the technique may be associated with considerable patient discomfort, together with the inconvenience of requiring repeated replacement/tightening of the cutting material.

A 'snug' rather than tight seton can effect a slower more gradual severance of tissue, and a silastic material may not only be more comfortable but also reduces the requirement for

replacement or retightening, as its elastic recoil allows a slow caudal migration [14]. The authors recommend the 'snug' seton in those patients with idiopathic fistulas and good preoperative function, with intersphincteric or transsphincteric tracks involving up to two-thirds of the sphincters below puborectalis (excluding anterior transsphincteric fistulas in women in whom more than one-third of the external sphincter is involved).

The Chemical Seton

An alternative to the traditional cutting seton, and one which has been used for many centuries in eastern parts of the world, is the so-called chemical seton, or Kshara sutra. This seton is a thread dipped in multiple layers of agents derived from plants which, apart from endowing antibacterial and anti-inflammatory properties, impart an alkaline (approximate pH 9.5) caustic nature that essentially burns through the enclosed tissues a rate of approximately 1 cm every 6 days [15].

Studies have compared the chemical seton with fistulotomy and shown no differences in rates of incontinence or recurrence rates, although the Kshara sutra is associated with prolonged healing and greater patient discomfort [16]. It is unknown whether such a technique has a role for the more problematic higher fistula.

Sphincter Preserving Techniques

Fistulectomy

Fistulectomy 'cores out' the fistula rather than dividing the tissue enclosed by its track. However, when compared to fistulotomy, there is little difference in recurrences rates, and healing is prolonged secondary to excess tissue loss.

The core-out technique has been advocated on the basis that the precise course of the track can be more accurately determined than by imaging or probing, thereby avoiding the potential creation of false tracks. Core out also reduces the risk of missing secondary tracks, which are seen as transected granulation tissue and thus can also be excised, and the relationship of the track to the sphincter complex can be more accurately ascertained [17].

The use of a new mechanical device, a 'fistulectome', has been described which cores out an approximately 2 mm circumferential thickness of the fistula tract [18]. However, the role of such a device in any fistula other than that with a single relatively straight track, and therefore unlikely to recur if treated by more traditional methods, is unclear.

The Loose Drainage Seton

The loose drainage seton can be used for different reasons in the management of anal fistulas [19]. A loosely tied thread can be used to drain sepsis, to allow subsidence of acute inflammation and either safer subsequent definitive surgery or as a long-term palliative measure aimed at symptom control (by preventing the fistula track from occluding and allowing sepsis to drain, thereby avoiding recurrent abscess formation). It can also be used as a marker to help determine the amount of muscle enclosed by the fistulous track, perhaps because scarring from previous surgery or relaxation under anaesthesia makes assessment difficult. In such circumstances, the proportion of enclosed sphincter may be more accurately determined when the patient is awake and the track marked by the thread.

Advancement Flaps

The use of advancement flaps to treat anal fistulas was first documented in 1912 by Elting [20]. He described two key principles: separation of the track from the communication with the bowel and adequate closure of that communication with eradication of all diseased tissue in the anorectal wall. At later dates others have added adequate flap vascularity, formation of a tension free flap, anastomosis of the flap to a site well distal to previous internal opening and resolution of any acute sepsis prior to definitive surgery [21]. There is little consensus as to whether the flap should be mucosal or include part if not all of the underlying internal sphincter. Mucosal advancement flaps seem to be associated with higher rates of fistula recurrence, whereas full or partial thickness advancement flaps can have an unpredictable effect on resting pressures and potentially severe functional consequences in the event of breakdown. The overall mean success rates

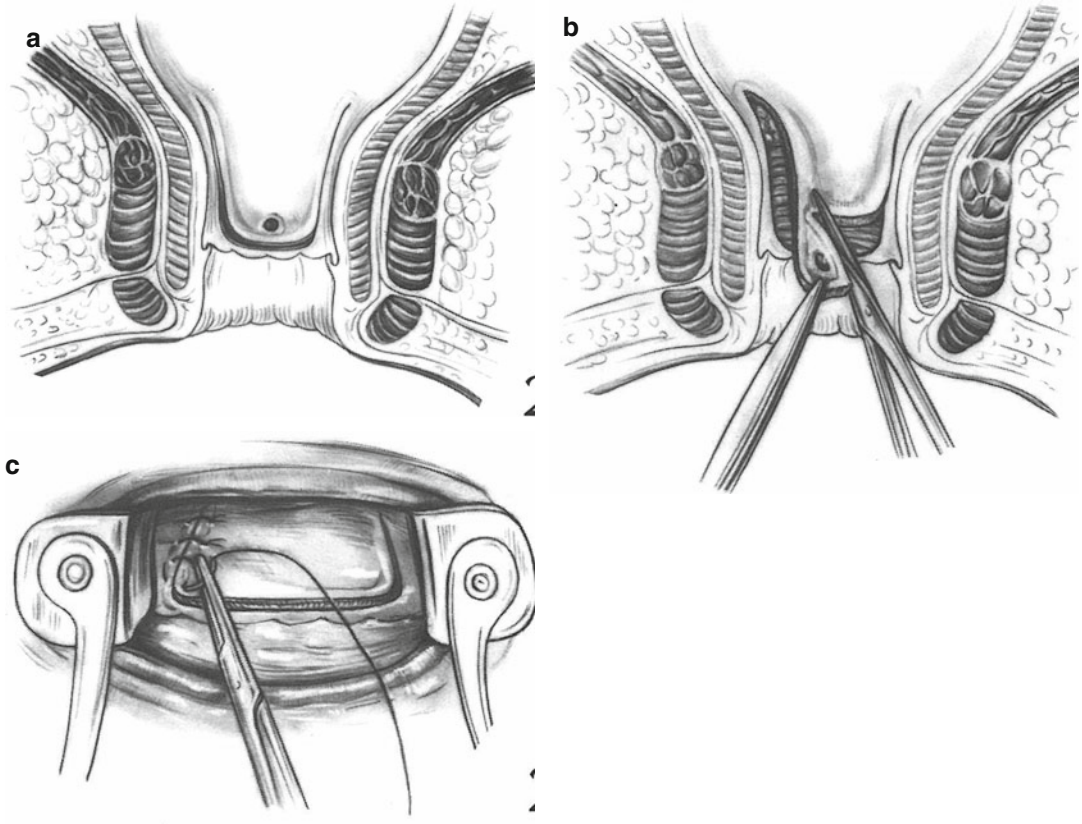

Fig. 19.2 (**a**–**c**) Illustrating the operative steps of an advancement flap (Reproduced from Hawley [26]. Reproduced by permission of Hodder Education

therefore probably lie between 50 and 60 %, and it seems that, as with all traditional surgical management strategies, the successful eradication of fistulas and the maintenance of continence remain directly competing variables. Additionally, advancement flaps are contraindicated in the presence of large internal openings (>2.5 cm), due to the risk of anastomotic breakdown, and a heavily scarred, indurated, woody perineum precludes adequate exposure and flap mobilisation (Fig. 19.2).

The Intersphincteric Approach

This technique was first described at St. Mark's Hospital in 1993 for use in high transsphincteric or suprasphincteric fistulas [22]. It was based on the centrality of chronic intersphincteric anal gland infection and the desire for sphincteric preservation. As originally described, the intersphincteric plane is entered, the fistula tract is excised in its entirety, the resultant openings in the external and internal sphincters are closed, and the internal and external openings are primarily closed. The procedure, however, was not widely adopted, perhaps because of its technically demanding nature. Almost two decades later, Rojanasakul described a simplified version of the technique (ligation of the intersphincteric tract, the 'LIFT' procedure), which has lead to a relative surge in uptake [23] (Fig. 19.3). The main areas of modification are ligation of the intersphincteric tract close to the internal opening, followed by excision of the intersphincteric component of the fistula only, removing the granulation tissue in the rest and suturing the defect at the external sphincter. As with all sphincter-preserving strategies, there are a wide range of fistula eradication rates reported (40–94 %) that generally decrease with the duration of follow-up.

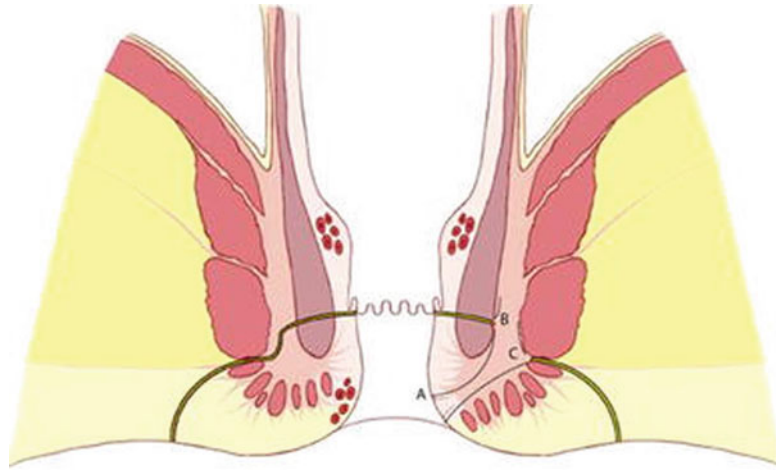

Fig. 19.3 Illustration showing the basic concept of the LIFT technique. (*A*) Approach via intersphincteric groove, (*B*) suture ligation of tract to close the internal opening and (*C*) suture ligation of defect in the external anal sphincter after removal of all infected granulation tissue (Reproduced from Rojanasakul [23], with permission from Springer)

Biological Materials

It seems that no matter how expertly chronic granulation tissue or epithelium lining the fistulous tract is removed, or the intersphincteric component of the fistula eradicated, or the tract separated from the anal canal contents (by mucosal advancement flap or simple stitch closure), these manoeuvres are no guarantor of successful fistula eradication. Proctologists have therefore started to look towards the rapidly developing worlds of biomaterials and tissue engineering to provide a material that bridges the defect left by excision of the track. The ideal biomaterial for this purpose would allow full host tissue incorporation and neovascularisation whilst withstanding premature degradation and bacterial colonisation. There are an increasing number of either biologically derived materials or synthetic materials designed to augment healing that are being marketed. The two most widely studied biomaterials for the treatment of anal fistulas are fibrin glue and a fistula plug composed of lyophilised porcine-derived small intestinal submucosa.

Fibrin glue is a tissue sealant that simulates physiological clot formation and was the first modern biomaterial to be used in the management of anal fistulas. It was initially received with great enthusiasm on account of its perceived benefits: the technique seemed simple to apply, repeatable (in that treatment failure did not compromise subsequent surgical options), spared the anal sphincter mechanism and avoided the prolonged discomfort associated with wound healing and repeated dressing changes. However, although early reports demonstrated excellent initial results, these were not replicated in the longer term. One of the main concerns with fibrin glue is that it simply does not exhibit the specific biological characteristics likely to be required for fistula healing, in that it does not allow host cell integration or neovascularisation to occur, and is degraded too rapidly to act as a scaffold for tissue repair [24]. Interestingly, fibrin glue does however encourage healing across its surface and therefore across the internal and external openings of the fistula within which it has been injected. At short-term clinical follow-up, this can mimic fistula healing [11].

In an attempt to overcome the biological failures of fibrin glue, the fistula plug was devised. A mesh, made of the same porcine-derived small intestinal submucosa, had demonstrated relative success in the treatment of hernias in potentially contaminated and contaminated wounds, and the material was therefore deemed to have an appropriate biological profile to heal anal fistulas. However, as with the glue, promising short-term results were offset by long-term fistula recurrence [25]. Both materials have now been reported to have widely variable success rates, with an overall mean clinical success rate of 50–60 %, although with no reported incidences of functional disturbance. These results mirror those seen with mucosal advancement

flaps. Technical factors, such as early implant extrusion, have been cited as one of the more consistent reasons for failure, and although this appears to be the case in the early post-operative period, it does not account for those whose treatment failed despite the material remaining in situ. The other factor that is most likely responsible for treatment failure in this latter group of patients is that the environment into which the materials are placed is not being optimised to facilitate healing (specifically, inadequate treatment of the primary track and failure of secondary track eradication). Interestingly, combining either of these two biomaterials with an advancement flap does not improve outcomes. A combination of the 'LIFT' procedure with insertion of a sheet of porcine-derived small intestinal submucosa may show promise and is currently enjoying popularity, though of course only time will tell if such a combination is effective in the long term.

As the understanding of anal fistula pathology, wound healing and the host response to materials (synthetic and biological) improves, so new biological sphincter-sparing strategies are being developed including the use of mesenchymal stem cells, autologous fibroblasts and xenogenic cross-linked collagen, the latter relatively resistant to premature degradation.

Conclusion

In conclusion, the following factors should be taken into account in the management of chronic anal sepsis:

1. Symptom severity
2. Sphincter function and integrity
3. Bowel habit
4. The fistula itself – aetiology, type, level and complexity
5. Patient expectation and honest counselling as to expected outcomes of the various techniques that may be applicable to that individual
6. A management strategy planned before any surgery

For these reasons, among others, it may be argued that the management of anal fistulas should be the domain of the appropriately trained and experienced proctologist, rather than the colorectal or general surgeon.

References

1. Lunniss PJ, Phillips RKS. Anal fistula: evaluation and management. In: Phillips RKS, editor. Colorectal surgery. 4th ed. Edinburgh: Elsevier Saunders; 2009. p. 223–43.
2. The NHS Information Centre, Hospital Episode Statistics for England. Inpatient statistics, 2009–2010. www.hscic.gov.uk/hes.
3. Sainio P. Fistula-in-ano in a defined population. Incidence and epidemiological aspects. Ann Chir Gynaecol. 1984;73(4):219–24.
4. Parks AG. The pathogenesis and treatment of fistula-in-ano. BMJ. 1961;18:463–9.
5. Parks AG, Gordon PH, Hardcastle JD. A classification of fistula-in-ano. Br J Surg. 1976;63:1–12.
6. Lunniss PJ, Phillips RK. Surgical assessment of acute anorectal sepsis is a better predictor of fistula than microbiological analysis. Br J Surg. 1994;81:368–9.
7. Eisenhammer S. The internal anal sphincter and the anorectal abscess. Surg Gynecol Obstet. 1956;103:501–6.
8. Malik AI, Nelson RL, Tou S. Incision and drainage of perianal abscess with or without treatment of anal fistula. Cochrane Database Syst Rev. 2010;(7): CD006827.
9. Choen S, Burnett S, Bartram CI, Nicholls RJ. Comparison between anal endosonography and digital examination in the evaluation of anal fistulae. Br J Surg. 1991;78:445–7.
10. Garcia-Aguilar J, Belmonte C, Wong WD, Goldberg SM, Madoff RD. Anal fistula surgery. Factors associated with recurrence and incontinence. Dis Colon Rectum. 1996;39:723–9.
11. Buchanan GN, Bartram CI, Phillips RK, Gould SW, Halligan S, Rockall TA, Sibbons P, Cohen RG. Efficacy of fibrin sealant in the management of complex anal fistula: a prospective trial. Dis Colon Rectum. 2003;46:1167–74.
12. Abcarian H. The 'lay open' technique. In: Phillips RKS, Lunniss PJ, editors. Anal fistula: surgical evaluation and management. London: Chapman & Hall Medical; 1996. p. 73–80.
13. Buchanan GN, Owen HA, Torkington J, Lunniss PJ, Nicholls RJ, Cohen CR. Long-term outcome following loose-seton technique for external sphincter preservation in complex anal fistula. Br J Surg. 2004; 91(4):476–80.
14. Hammond TM, Knowles CH, Porrett T, Lunniss PJ. The Snug Seton: short and medium term results of slow fistulotomy for idiopathic anal fistulae. Colorectal Dis. 2006;8:328–37.

15. Deshpande PJ, Sharma KR. Successful non-operative treatment of high rectal fistula. Am J Proctol. 1976;27(1):39–47.
16. Ho KS, Tsang C, Seow-Choen F, Ho YH, Tang CL, Heah SM, et al. Prospective randomised trial comparing ayurvedic cutting seton and fistulotomy for low fistula-in-ano. Tech Coloproctol. 2001;5:137–41.
17. Lewis A. Core out. In: Lunniss PJ, Phillips RKS, editors. Anal fistula. Surgical evaluation and management. London: Chapman & Hall Medical; 1996. p. 81–6.
18. Tasci I. The fistulectome: a new device for treatment of complex anal fistulas by "core-out" fistulectomy. Dis Colon Rectum. 2003;46:1566–71.
19. Williams JG, MacLeod CA, Rothenberger DA, Goldberg SM. Seton treatment of high anal fistulae. Br J Surg. 1991;78:1159–61.
20. Elting AW. X. The treatment of fistula in ano: with especial reference to the whitehead operation. Ann Surg. 1912;56:744–52.
21. Lunniss PJ. The role of the advancement flap technique. Seminars Colon Rectal Surg. 1998;9:192–7.
22. Matos D, Lunniss PJ, Phillips RK. Total sphincter conservation in high fistula in ano: results of a new approach. Br J Surg. 1993;80:802–4.
23. Rojanasakul A. LIFT procedure: a simplified technique for fistula-in-ano. Tech Coloproctol. 2009;13: 237–40.
24. Hammond TM, Grahn MF, Lunniss PJ. Fibrin glue in the management of anal fistulae. Colorectal Dis. 2004;6:308–19.
25. Garg P, Song J, Bhatia A, Kalia H, Menon GR. The efficacy of anal fistula plug in fistula-in-ano: a systematic review. Colorectal Dis. 2010;12:965–70.
26. Hawley P. Low anal fistula. In: Fielding LP, Goldberg SM, editors. Rob & Smith's operative surgery of the colon, rectum and anus. Oxford: Butterworth-Heinemann; 1993. p. 826.

Perianal Skin Conditions

20

Tamzin Cuming, Angela C. Bailey,
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Introduction

Patients with conditions affecting the perianal skin present to various clinics, from proctology to sexual health to general practice. The boundaries of such specialties as dermatology, infectious disease and gastrointestinal surgery are crossed by the range of diagnoses that underlie complaints in the perianal region. The problem may be local to the anus, be an indication of intestinal disease or represent a manifestation of a generalized dermatological disorder. In the latter case, it is notable that the moist and warm environment of the perianal region often produces differing appearances to those characteristic of the disease elsewhere.

The perianal region is a common site for presentation of sexually transmitted infections, which are addressed in detail.

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Perianal Anatomy and Function

The perianal skin is the circumferential area of skin outside the anal verge, the border with the anal canal. The anal canal develops embryologically from the posterior cloacal membrane which unites hindgut and proctodeum by rupturing at 7 weeks' gestation. Within it, a transition occurs in cell type that reflects the change from ectodermal to endodermal origin. The perianal skin therefore, with the distal anus, derives from the proctodeum and is supplied by the pudendal vessels with lymphatic drainage to the inguinal lymph nodes. Sensation is somatic via the pudendal nerve (S2, S3, S4), and the perianal area has its own dermatome (S4) with an additional, posterior area towards the coccyx supplied by S5, see chapter.

Perianal skin has an inner circumferential area closest to the anal verge with characteristic corrugation brought about by the insertion of the intersphincteric longitudinal fibres into the dermis of the skin. This inner area of skin extends approximately 10–15 mm from the anal verge – equating to the diameter of the underlying external anal sphincter [1]; it differs from the rest of the perianal skin in three ways: it is more pigmented, it has only fine hairs and the corrugation allows great stretch.

Microscopically, the structure of the perianal skin is that of skin elsewhere. The perianal dermis is particularly adapted to stretch with many thick elastin fibres and is well supplied with

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adnexa: the eccrine glands with sudomotor sympathetic innervation, pilosebaceous units and apocrine glands.

The dermal-epidermal border is bounded by the basal layer of undifferentiated cells in an undulating rete peg shape which produces keratinocytes that mature and differentiate as they move up through the three layers that constitute the epidermis: the stratum spinosum, the stratum granulosum and the stratum corneum.

The perianal skin thus comprises keratinized, stratified squamous epithelium with plentiful skin appendages and is exquisitely sensitive to pain. At the anal verge, the skin changes to the non-keratinized, stratified squamous epithelium without skin appendages, but still with somatic innervation and sensitivity to pain, which continues proximally until the transition zone within the anal canal (see Anatomy chapter).

Normal Flora

The perianal skin is an intertriginous (flexural) area with apposition of skin surfaces where warmth and moisture encourage the proliferation of bacteria and fungi to a high level. The anal continence mechanism normally prevents mucus and faeces from constantly bathing the perianal skin; however, there is regular exposure to gastrointestinal as well as skin flora. The potential pathogens *Staphylococcus aureus* and *Candida* species are common commensals as is *Staphylococcus epidermidis* of skin origin and *Bacteroides* species, *Enterococcus faecalis*, and Enterobacteriaceae of the lower gastrointestinal tract. Fungal colonization with *Candida albicans* is commonly asymptomatic [2].

Dermatology for the Proctologist

Terminology

Dermatological terminology is employed to describe the appearance skin changes in a standardized and structured manner (see Table 20.1).

Table 20.1 Dermatological terms

Some commonly used dermatological terms
Macule – a flat skin lesion <5 mm in diameter
Patch – a flat skin lesion >5 mm in diameter
Papule – a raised skin lesion <5 mm in diameter
Plaque – a raised skin lesion >5 mm in diameter
Nodule – a solid raised skin lesion that extends into the dermis or subcutaneous tissue
Erosion – partial loss of the epidermis
Ulcer – full-thickness loss of the epidermis
Vesicle – a fluid-filled blister <5 mm in diameter
Bulla – a fluid-filled blister >5 mm in diameter
Pustule – a pus-filled blister
Xerosis– dry skin
Telangiectasia – permanent dilation of small blood vessels in the skin
Koebnerization – appearance of rash/lesions of the same morphology at sites of trauma
Lichenification – thickening of the skin due to repeated rubbing

History

With a careful dermatological history (Box 20.1) and where relevant, a sexual history (Box 20.2), a differential diagnosis can often be deduced with clinical assessment alone (Table 20.2).

Box 20.1 Dermatological History

- Presenting symptoms
  - Itching – especially at night
  - Bleeding
  - Pain/soreness (e.g. on defecation, constant/intermittent, generalized/localized sharp/dull)
  - Lumps (e.g. warts)
  - Concerns of patients re symptoms (especially anxiety and embarrassment)
- Past medical history, for example, Crohn's disease, atopy and HIV
- Family history of skin disease, for example, psoriasis and atopic dermatitis
- Drug history including topical medication
- General habits including use of soaps, shower gel, baby wipes and deodorants

Box 20.2 Tips on Taking a Sexual History

Be relaxed and ask questions as you would normally	
Be non-judgmental	
Prepare:	
Request that accompanying persons leave for a moment	
Explain to the patient that you need to ask some personal questions about their sex life as it may be related to their symptoms	
Reassure them that the information will be kept confidential	
Obtain permission to proceed	
Suggested questions:	**Why ask this?**
1. When was the last time you had sex?	Find out if sexually active and open conversation
(a) Was that a regular partner or a casual partner?	Gives an idea about risk taking
(b) Was the partner male or female?	Ascertain sexual orientation; risk of STIs
(c) Is the patient or the partner from another country?	Risk assessment for STIs
(d) With a condom?	Risk taking behaviour
2. When did you last have sex with a different person	Rate of partner change is an important risk factor for STIs
Clarify details as above	
3. Have they ever had anal sex?	Important risk factor for perianal STIs
If so, was a condom used?	
4. For men: have you ever had sex with a man?	This may affect risk of HIV, hepatitis B, even if long ago
5. Have you ever injected drugs or had a blood transfusion (especially in a developing country or many years ago)?	Risk for blood-borne viruses
In all sexually active or intravenous drug-using (past or present) persons, a blood-borne virus (HIV, hepatitis B and C) and syphilis screen should be offered. Ask if they have been previously tested and if so when and what the results were	

Table 20.2 Differential diagnoses

The multifactorial nature of perianal complaints can combine to produce a large differential diagnosis for any particular symptom. This box is a guide to possible diagnoses to consider
White patches
Vitiligo
Lichen sclerosus
AIN (see premalignant conditions)
Lichen simplex
Red lesions
Dermatitis/eczema
Atopic
Contact
Irritant/chemical
Allergic
Seborrhoeic
Psoriasis
Infection – bacterial or fungal
Vascular tumours, e.g. hemangioma
Fissures of the skin
Crohn's (multiple, ± anal canal fissure)
Psoriasis
Dermatitis
Herpes simplex virus (HSV)
Itching – see Pruritus Ani Chap. 21
Ulcer
Herpes (HSV)
Syphilis
Lymphogranuloma venereum (LGV)
Human immunodeficiency virus (HIV)
Malignancy (see Malignant Disease)
Crohn's disease
Varicella zoster
Cytomegalovirus (CMV)
Nicorandil
Behçet's disease
Trauma/factitious
Deep mycoses
Chancroid
Donovanosis (granuloma inguinale)
Blistering disorders
Vesicles
Varicella including varicella-zoster virus
Herpes simplex
Stevens-Johnson syndrome
Bullae
Pemphigus vulgaris
Bullous drug eruption, e.g. fixed drug eruption

(continued)

Table 20.2 (continued)

Lumps
Lipoma
Epidermoid cyst
Dermoid cyst/teratoma
Warts (condylomata acuminata)
Molluscum contagiosum
Condylomata lata
Seborrhoeic keratoses
AIN/bowenoid papulosis
Pseudofolliculitis (ingrowing hairs)
Prominent skin folds

Examination

After inspection of the genital and perianal areas, digital and speculum examination of the vagina and sigmoidoscopy of the rectum should be considered. Examination of the scalp, oral cavity, hair and nails and wider cutaneous examination can then be targeted according to the findings, for example, the extensor surfaces of limbs in psoriasis and oral cavity for lesions in secondary syphilis.

Diagnosis

The perianal skin is an accessible site for tissue sampling if a diagnosis has not been arrived at using clinical assessment. Skin scrapings (for microscopy and culture), swabs and biopsy are the mainstays of investigation.

For the proctologist, it is important to consider viral swabs for herpes simplex virus as these are often not part of the normal surgical management (see later in this chapter).

Treatment

Simple treatments, such as the use of emollients as soap substitutes, moisturizers and barrier creams to protect the skin are highly effective general measures in the management of many skin conditions. These agents are particularly useful in the management of dry skin, pruritus, psoriasis and irritant and atopic dermatitis (eczema).

Local anaesthetics have a role in painful disorders such as fissure in ano and first episode of herpes simplex virus infection (HSV) and are generally well tolerated, although contact dermatitis has been described [3].

Antihistamines are used orally for pruritic lesions.

Antimicrobial agents have an important role. Many of these agents possess anti-inflammatory activity and are used to treat both infectious and non-infectious skin disease, for example, the use of tetracycline in hidradenitis suppurativa.

Antifungals are used in managing dermatophytosis (ring worm), candidiasis (thrush), subcutaneous and systemic fungal infections and in controlling malassezia yeasts in seborrhoeic dermatitis.

Topical corticosteroids are the mainstay of the management of allergic and inflammatory dermatological conditions. There are low, intermediate and high potency topical steroid preparations. Weak preparations can be bought over the counter (Box 20.3).

Chronic use of potent topical corticosteroids can lead to atrophy and telangiectasia. Therefore, it is preferable that long-term management of patients requiring such agents be carried out under medical supervision. With purely perianal use, topical steroids result in insignificant systemic absorption.

Steroid-sparing agents include calcineurin inhibitors (e.g. pimecrolimus, tacrolimus). They should be used with caution and with specialist medical advice due to concerns regarding possible carcinogenic effects with long-term use.

Tumour necrosis factor (TNF) alpha inhibitor drugs (e.g. infliximab, adalimumab) are effective in Crohn's disease [4], and there is limited evidence for their use in hidradenitis suppurativa [5].

Other treatment options include imiquimod, retinoids and laser. Surgical management is necessary for abscesses, biopsy and excision.

Sexual Health for the Proctologist

Sexually transmitted infections (STIs) frequently involve the perianal area. Sexual practices involving the anus are common: anal intercourse in the last year was reported by more than 10 % of UK

Box 20.3 Topical Corticosteroid Preparation Potencies

Mild:
Hydrocortisone 0.5 %; 1 %
With antifungals:
Hydrocortisone 1 % + clotrimazole 1 %
Hydrocortisone 1 % + miconazole 1 %
With antibacterials:
Hydrocortisone 1 % + fusidic acid 2 %
Moderate:
Clobetasone butyrate 0.05 %
With antimicrobials:
Clobetasone butyrate 0.05 % + oxytetracycline 3 % + nystatin 10,000 units/g
Potent:
Betamethasone valerate 0.1 %
Mometasone furoate 0.1 %
Hydrocortisone butyrate 0.1 %
With antimicrobials:
Betamethasone valerate 0.1 % + fusidic acid 2 %
Very potent:
Clobetasol propionate 0.05 %
Diflucortolone valerate 0.3 %
Topical antibacterial preparation:
Fusidic acid 2 %
Mupirocin 2 %
Topical antifungal preparation:
Clotrimazole 1 %
Miconazole 2 %
Terbinafine 1 %

adults and more than 20 % of US adults in household surveys [6, 7]. Infections are discussed individually, but the following principles apply to management of all STIs:

1. Take a sexual history (see Box 20.2).
2. Confirm the diagnosis in the laboratory if at all possible. Ensure clinics and theatres are supplied with the equipment for viral and Chlamydia (nucleic acid amplification tests – NAATs) testing.
3. Treat diagnosed infections quickly (e.g. herpes simplex infection) or refer rapidly for urgent treatment (e.g. for syphilis), recommending sexual abstinence in the meantime. Ensure local guidelines are followed.
4. STIs travel in packs – always screen for other infections or recommend attendance elsewhere for screening, including HIV, if you suspect or diagnose one STI.
5. Management of STIs is not complete without partner screening and treatment. Refer for management of partners or at least ensure the patient is aware that their partners are at risk.
6. STI diagnoses are associated with stigma and psychological morbidity. Some patients may require referral for specialist support.

Non-infectious Skin Disorders

Dermatitis (Eczema)

Dermatitis is a general term to describe inflammation of the skin. It is often used synonymously with eczema, a term that derives from the Greek 'to boil'. It describes a group of diseases which present with an itchy red rash that may show oozing, crusting or scaling.

Atopic Dermatitis (Atopic Eczema)

Atopic dermatitis/eczema is a pruritic eruption that is recurrent, usually flexural in adults, symmetrical and associated with a predisposition to atopy (personal or family history of asthma, eczema or hay fever). It can present in infancy, childhood or adulthood. Perianal involvement can rarely occur in all three stages. The napkin area is usually spared in infancy but can be affected [6]. Atopic dermatitis accounts for 3–5 % of cases of pruritus ani in adults [8]. Although atopic dermatitis is common, isolated involvement of the perianal area is rare.

Treatment involves the use of antihistamines to control itching and mild to moderately potent topical corticosteroid preparations to control inflammation.

Contact Dermatitis

Contact dermatitis is a common underlying or contributory cause of perianal symptoms and

presents as an erythematous lesion representing a reaction to a substance which has breached the skin barrier [9].

Two forms of contact dermatitis are described which can be difficult to distinguish clinically: irritant contact dermatitis, where the cells are directly damaged by the agent in question, and allergic contact dermatitis, where such a substance induces a delayed-type hypersensitivity reaction. Predisposing factors to this multifactorial condition include atopic dermatitis, genetic factors and environmental cofactors such as moisture and occlusion [10].

Irritant Contact Dermatitis

This is caused by either exposure to weak irritants (e.g. soap, shower gel) or contact with strong irritants such as acids and alkalis and is a nonimmune-mediated inflammatory skin reaction presenting in acute or chronic forms.

Symptoms include irritation and soreness or a burning sensation. The skin usually appears erythematous with vesicles in the acute stage with scaling, fissures and lichenification in the chronic stage. Potential irritants in the perianal area include hygiene products, wet wipes, detergents, topical haemorrhoid preparations and products for the relief of itching which can paradoxically contain irritants as preservatives. Mucus and faecal matter from incontinence, prolapse of haemorrhoids, or from the rectal mucosa lead directly to irritant contact dermatitis, as it does in infants (diaper/nappy rash).

Treatment involves withdrawal of the irritant or protection against it with barrier creams along with application of weak or moderately potent topical corticosteroid cream or ointment.

Allergic Contact Dermatitis

Allergic contact dermatitis can be difficult to distinguish from irritant contact dermatitis. It is due to a delayed hypersensitivity reaction (type IV) [11]. An analysis of 1,168 suspected anogenital contact dermatitis patients showed that allergy was responsible in 25 %, with the main culprits being the patient's own products as well as lanolin and fragrances, and hence, these should be used for allergen patch testing which is now widely available [12]. Nail varnish allergy has been described as a cause of perianal rash [13].

A preservative used in the increasingly popular moist toilet tissues/baby wipes (methylchloroisothiazolinone/methylisothiazolinone, MCI/MI) is known to cause allergic contact dermatitis [14]. Ingested allergens such as nickel [9] or spices in food, particularly those related to balsam of Peru (cinnamon, cloves, nutmeg, vanilla), can lead to perianal dermatitis [15]. More commonly, however, allergic contact dermatitis is due to preparations intended to ameliorate anal symptoms: haemorrhoid creams, deodorants and local anaesthetics [16]. Treatment involves identifying and removing contact with the allergen to which the patient is sensitized and use of mild to moderately potent topical corticosteroid preparations to control inflammation.

Seborrhoeic Dermatitis (Seborrhoeic Eczema, Cradle Cap)

Other affected sites: scalp, eyebrows, retroauricular and genitocrural.

Seborrhoeic dermatitis is a chronic inflammatory condition which can affect the perianal area. It is relatively common (3–5 %) and more prevalent in men. It is characterized by pruritic erythematous areas with greasy yellowish fine scaling located in areas of high sebum production, such as the scalp, back, groins, chest and the genitocrural region. Scaling may be less obvious in the perianal area, and examination of other typical sites will give a clue to the diagnosis.

Seborrhoeic dermatitis is etiologically associated with the yeast *Malassezia furfur* (*Pityrosporum ovale*), and the two ages of greatest incidence – infantile (2 weeks to 12 months) and adolescent-adult – correspond to periods of increased sebum production. However, the relationship is not simple. Treatment involves controlling seborrhoea of the scalp with antifungal shampoos such as ketoconazole or zinc pyrithione

at the same time as low to intermediate potency topical steroids in combination with antifungals for the perianal skin lesions. Severe or treatment-resistant seborrhoeic dermatitis may require oral antifungals and is an indicator condition for unrecognized human immunodeficiency virus (HIV) infection [17].

Secondary bacterial infection can occur, and the differential diagnosis includes flexural psoriasis and intertrigo.

Lichen Simplex Chronicus (LSC)

This is a chronic eczematous condition characterized by localized plaques due to repeated rubbing and scratching in response to pruritic stimuli. The skin, initially erythematous, becomes lichenified. This leads to further itching which results in habitual rubbing and more lichenification, the 'itch-scratch cycle'. Any area that can be conveniently reached can be affected including the perianal area where lesions can be asymmetrical (see Fig. 20.1). Precipitating factors include heat, sweat, chemical irritation and 'psychological'. Twenty-five percent of patients have a background of atopy [18, 19].

LSC must be distinguished from psoriasis, contact dermatitis and anal intraepithelial neoplasia (AIN). All causes of pruritus ani need to be considered in the search for an underlying trigger, including candidiasis, dermatophyte infection, parasitic infestation as well as faecal incontinence, haemorrhoids and neoplasia (see Chap. 21). Topical steroids are the mainstay of treatment. Sedative antihistamines are helpful for controlling pruritus at night. Other measures include generic pruritus advice: avoiding soaps, using cotton anal plugs and barrier creams. Patients should be made aware that the rash will not resolve until scratching and rubbing are stopped.

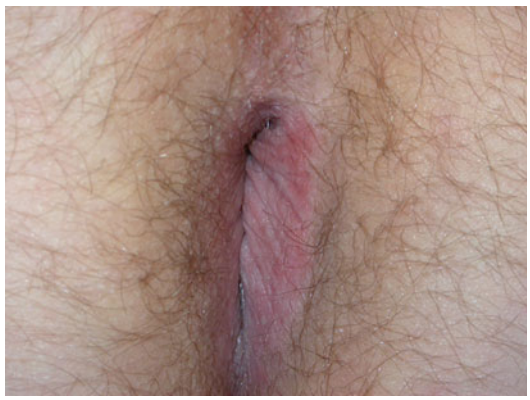

Fig. 20.1 Lichen simplex chronicus. Note lichenification over the perianal area especially on the left side (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

Psoriasis (Inverse Psoriasis, Flexural Psoriasis)

Associated sites: extensor surfaces especially elbows, knees, scalp and nails.

Psoriasis is a chronic inflammatory disorder, which, in its most common form, is characterized by erythematous scaly papules and plaques typically seen over scalp and extensor surfaces of the body. These are generally asymptomatic but lesions occasionally can be pruritic.

Psoriasis affects 1–5 % of the population in Western societies, with bimodal peaks in the early 20s and mid-50s, with men and women equally affected. There are several types of psoriasis, but the type seen in the perianal area is the 'flexural' or 'inverse' type which also affects axillae, popliteal fossae and inguinal and intergluteal areas, as well as the inframammary area in women.

Well-defined erythematous plaques or patches with fissuring are seen which differ from psoriasis of non-intertriginous areas due to minimal scaling (see Fig. 20.2). Genital and anal lesions usually occur concomitantly. Although isolated perianal psoriasis can rarely occur, other sites are often affected and should be examined. Nail changes include irregular pits, onycholysis (separation of nail from the nail bed), subungual hyperkeratosis (accumulation of keratin material underneath the nail) and translucent yellowish spots under the nail plate (oil drop sign).

Perianal psoriasis can mimic dermatitis, fungal or bacterial infection due to the lack of characteristic scaling at this site.

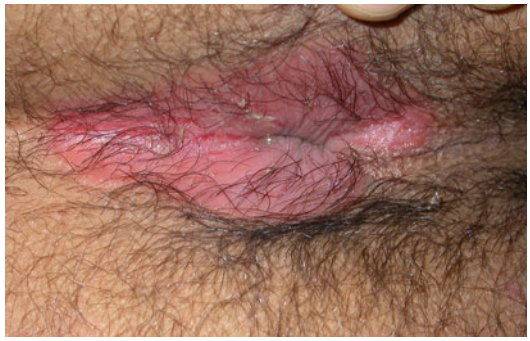

Fig. 20.2 Psoriasis. Well-defined erythematous plaque with fissuring. Scaling is usually not a feature at flexures. (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

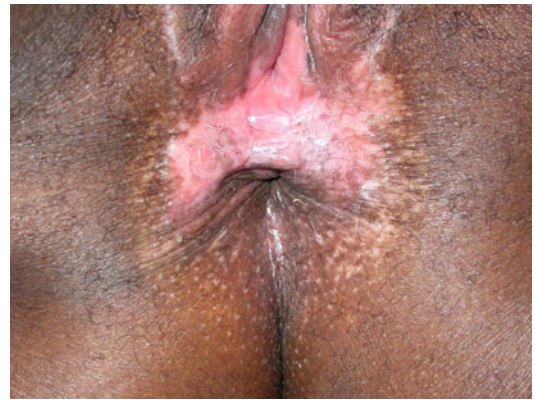

Fig. 20.3 Lichen sclerosus. Note whitish sclerotic area over perineum extending to the perianal area and hypopigmented macules and papules confined to the rest of the perianal area. The vulva was also affected (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

Psoriasis may be the initial presentation of HIV infection, or HIV can aggravate existing psoriasis (see later in this chapter).

Treatment is with mild or moderately potent topical steroids with or without antifungal agents. Vitamin D analogues can be used as steroid-sparing agents.

Lichen Sclerosus (Lichen Sclerosus et Atrophicus and Balanitis Xerotic Obliterans, LS)

Additional site: vulva.

Lichen sclerosus is a dermatological disorder of unknown aetiology with predilection for the anogenital region. There is a peak of incidence in childhood. In adulthood, women are affected more than men (10:1), usually at a postmenopausal stage.

LS is considered an autoimmune disorder with a similar profile to vitiligo, morphea and autoimmune disease of the thyroid [20].

It is characterized by ivory-white papules that become confluent to form whitish plaques that subsequently become atrophic (see Fig. 20.3). Keratotic follicular plugs may be seen on the surface in early lesions. Focal areas of petechiae or purpura may be seen in established lesions.

In women, involvement of the vulva and perianal area simultaneously gives rise to a 'figure of eight' or 'hour glass' appearance. Scarring develops subsequently. Malignancy can occur in 3–4 % of cases of lichen sclerosus and needs to be excluded if plaques or ulcerated lesions develop.

Biopsy helps to confirm the diagnosis and exclude premalignant and malignant changes. The mainstay of treatment is potent topical steroid creams. Long-term follow-up is recommended in view of the potential for malignant change.

Lichen Planus (LP)

Additional sites: oral mucosa, trunk and nails.

Lichen planus is a chronic inflammatory pruritic mucocutaneous disease. The classic lesion is an erythematous flat-topped violaceous papule. Whitish reticular lines can be seen on the surface (Wickham's striae). Koebner phenomenon can occur with new lesions developing at sites of injury. Whitish reticulated patches may be seen in the oral cavity, and nail changes include longitudinal ridging and pterygium (fusion of the nail fold to the matrix).

There are several types of LP. The papular and hypertrophic types can affect the natal cleft and buttocks.

The erosive type of LP affects the anogenital area and the oral cavity. It is painful due to the desquamation, is chronic and is recalcitrant to

treatment. There is also a small (2–3 %) risk of developing squamous cell carcinoma.

Lichen planus usually responds to potent topical steroids. Tacrolimus or pimecrolimus have also been used with some success [21]. Refractory cases may require oral immunosuppression. Late presentation of erosive lichen planus can cause adhesions and lead to stenosis of the introitus or, rarely, of the anus.

Hidradenitis Suppurativa (Acne Inversa, Verneuil's Disease, HS)

Associated sites: inguinal, axillae, inframammary and buttocks.

Hidradenitis suppurativa is defined as a 'chronic, inflammatory, recurrent, debilitating skin disease' [22]. It is a cause of chronic abscesses and sinus formation in apocrine gland-bearing areas of the skin – most commonly the axillae, groins, perianal and buttock areas.

HS is extremely rare before puberty and more common in women (3:1). Involvement of axillary, groin and inframammary areas is seen more in females, whereas the perianal region and buttocks are more affected in men. Around 25 % have a familial form with an autosomal dominant pattern [23].

The exact pathogenesis of HS is not known, but it is thought to be due to occlusion of the pilosebaceous-apocrine units which leads to rupture of the follicle and an aseptic chronic inflammatory process resulting in painful deep-seated red nodules and sinus formation [22].

There is an association with other follicular occlusion disorders leading to the concept of a tetrad which also includes pilonidal sinus, acne conglobata and dissecting cellulitis [24]. Other predisposing factors include smoking, obesity and altered sensitivity to hormones particularly androgens although their influence has yet to be fully elucidated [25].

The role of bacteria is debated: secondary infection may occur; a primary causal role is not established.

The clinical diagnostic hallmark is the presence of double or multiple comedones ('tombstone' comedones). The nodules do not point which distinguishes them clinically from bacterial furunculosis. The Hurley classification consists of three stages: stage one constitutes abscess formation and is the most common type (70 % of cases of HS); stage two involves sinus formation and cicatrisation (25 %); and 1 % progress to stage three severe disease with diffuse-bridged scarring and interconnecting tracts [26].

The differential diagnosis includes Crohn's disease, furunculosis, infected epidermoid cysts and relatively rare infections such as tuberculosis, lymphogranuloma venereum, nocardia and actinomycosis.

Differential from cryptoglandular fistulation is usually made by inspection: HS is a superficial disease which although occasionally involving the anal canal does not fistulate across or within the sphincters [27] (see Fistula-in-Ano chapter). Despite this, the differential diagnosis can be difficult clinically and require both examination under anaesthetic and MRI scan of the perineum to exclude deep extension, as well as biopsy.

Diagnosis depends on the triad of features: typical clinical appearance, typical sites and the recurrent nature of the disease.

Conservative management includes general measures such as reducing heat, humidity and sweating with loose-fitting undergarments, local antiseptic washes, losing weight and stopping smoking.

Antibiotics are used and may be needed long term. The most effective are those with an anti-inflammatory function, such as the tetracyclines, clindamycin and rifampicin. Modulation of the sex hormones with anti-androgens is employed, for example, with the use of oral contraceptives containing cyproterone acetate. Retinoids also have been used therapeutically as have corticosteroids, zinc gluconate [28] and topical resorcinol treatment [29]. Nd: YAG laser therapy has found a place [30]. The role of TNF-alpha inhibitors is being explored: response in Hurley stage III can be rapid with a dramatic effect [31]. Surgery is used in three main ways: incision and drainage of abscesses, deroofing and wide excision. Surgery in combination with TNF-alpha inhibitors may be required for severe disease (see Fig. 20.4).

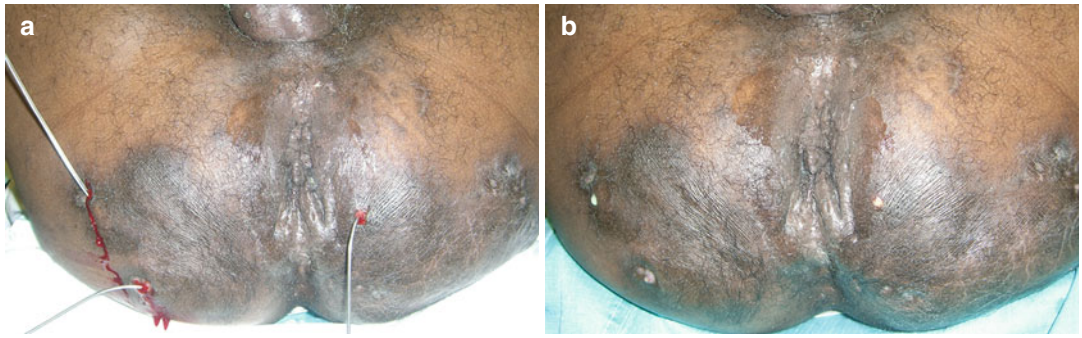

Fig. 20.4 Hidradenitis suppurativa: superficial fistulation and sinus tracts, (**a**) multiple fistulae of perianal hidradenitis suppurativa (**b**) probes in sinus tracks, perianal hidradenitis suppurativa (Courtesy of Mr P A Giordano, Whipps Cross University Hospital, London)

Crohn's Disease (Crohn's Disease, Terminal Ileitis)

Associated sites: ileum, colon, oral cavity and extra-intestinal.

It is recognised that the gastrointestinal tract from mouth to anus can be affected with the characteristic full-thickness non-caseating granulomatous inflammation of Crohn's disease. There are extra-intestinal manifestations such as the rare cutaneous involvement with granulomatous inflammation (metastatic Crohn's disease) as well as more commonly associated joint, eye, skin and hepatobiliary disorders.

Perianal Crohn's disease represents a major part of Crohn's morbidity, with pain and incontinence being common features. Perianal involvement in Crohn's disease is fairly common, with 25 % of those with terminal ileal and 75 % of those with colonic Crohn's affected, and the more distal the affected Crohn's segment, the more likely the perianal involvement.

Crohn's disease can affect ages from childhood onwards; however, it has a bimodal distribution with a peak at age 20–40 years and a second at ages 60–70 years. Crohn's patients with perianal disease present 8 years younger than those without [32]; women are slightly more likely to have an associated perianal diagnosis (57 %) [33]; perianal disease at an early age is associated with a more severe course for both luminal and perianal Crohn's [32, 34]. It is a risk factor for proctectomy and stoma formation [33, 35].

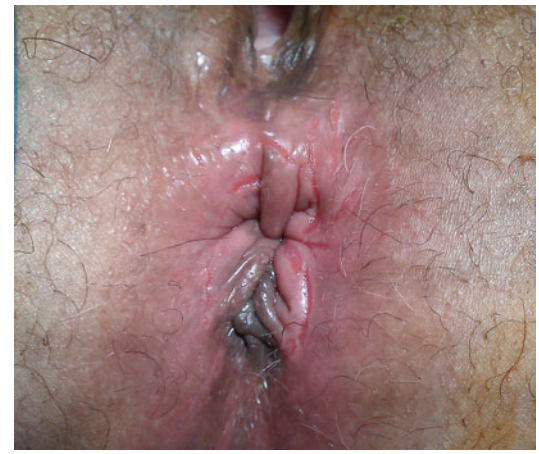

Fig. 20.5 Crohn's disease of the perianal area without fistulation: skin tags and superficial fissuring (Courtesy of Mr P A Giordano, Whipps Cross University Hospital, London)

Perianal Crohn's can predate the appearance of Crohn's disease elsewhere which it behoves the clinician to consider [36] (see Fig. 20.5).

A number of scoring systems have been developed: the Cardiff and Perianal Crohn's Disease Activity Index being used mainly in research [37, 38]. Psychological morbidity of Crohn's disease is disproportionately focused on the activity of the perianal component of the disease [39].

Skin Tags

Skin tags are the most common perianal feature, present in 40 % [37]. Unlike non-Crohn's skin tags, those due to Crohn's can be painful. There are two types of tag. Type 1 are inflammatory in

nature, enlarge and swell and can be purplish or cyanotic, hard and tender, and their excision is contraindicated due to complications with wound healing [35]. Type 2 skin tags are flesh coloured, wide based and soft ('elephant ears'), with no intermittent swelling or associated pain; they are more prevalent and larger but essentially similar in nature to those occurring in the population at large and are amenable to surgical excision.

Fistulae

Fistulating Crohn's disease is the first presentation of the disease in around 10 % of patients, and complex fistulae carry a 50 % proctectomy risk [40] (see Fistula chapter).

Fissures

Anal fissures in Crohn's disease can precede the development of intestinal symptoms of the disease and occur in 20 % of patients [41]. Fissures that are multiple and in unusual sites are classically due to Crohn's disease; however, two-thirds of Crohn's fissures are located in the same posterior midline position as is typical of idiopathic fissure [42] (see Fissure chapter). The distinction from idiopathic fissure is of paramount importance as the priority in Crohn's fissure management is the preservation of sphincter function, particularly in women. Conservative management has a 75 % success rate [43]. Crohn's fissure in ano, though often less painful than those of idiopathic origin, can be persistent and recurrent over many years.

Abscesses

Perianal abscesses in Crohn's disease are common (60 %) and can also be a first presentation of the disease. Seventy percent are associated with a transsphincteric fistula [40]. Prompt management with incision and drainage to control the sepsis will limit the progression and potential risk of septic damage to sphincter muscles (see chapters on Sepsis/Fistula).

Ulcers

Ulcers in cutaneous Crohn's disease range from superficial fissuring (which should be distinguished from psoriasis and dermatitis) to frank and deep ulcers with undermined edges and cavitation and unremitting pain. Linear ulcers in a 'knife-cut pattern' may be seen. Attempts have been made to distinguish between superficial ulceration with good prognosis and more significant cavitating ulceration in Crohn's disease of the perineum [35, 37]. Ulceration can predate diagnosis of Crohn's disease or any intestinal symptoms by some years.

The prognosis of the ulceration is related to associated fibrosis and stricture formation (see below). It may represent an indication of severe disease [44]. Patients with Crohn's disease and significant ulceration have at least a 25 % risk of proctectomy; however, this may primarily be related to stenosis as a consequence of ulceration [33]. 50 % of the perianal ulcers in Crohn's eventually heal spontaneously although approximately 20 % persist [44].

Metastatic Crohn's Disease

This is a very rare cutaneous manifestation of Crohn's disease defined as affecting an area of skin not contiguous with the gastrointestinal tract. Case reports have shown a predilection for the perianal area as well as the legs, with non-caseating granulomas being the defining histological feature [45].

Summary

Patients with perianal Crohn's disease have significant morbidity from loss of elasticity of the anus, pain from fissuring, tender skin tags and septic consequences of fistulae, as well as from anal canal and rectal disease. Ultimately defunctioning, or proctectomy may be required, and incontinence is a risk. Therefore, conservative management even of minor lesions is recommended if at all possible from their first presentation.

Hidradenitis Suppurativa and Crohn's Disease

Crohn's disease and hidradenitis suppurativa may be associated. Twenty-four of a Cleveland clinic cohort of 61 hidradenitis suppurativa patients were found to have coexistent Crohn's disease [46]. HS may precede or complicate Crohn's

disease. The two disease entities may be linked by immune dysregulation, and both are worsened in smokers. This association has led to the use of anti-TNF-alpha agents in the management of hidradenitis suppurativa [5].

Behçet's Disease

Associated sites: eyes and mouth.

Behçet's is a multisystem vasculitic disorder featuring recurrent mucocutaneous and genital ulceration with eye involvement and cutaneous lesions ranging from pustules to erythema nodosum. Gastrointestinal manifestations include ulcers that may occur anywhere from the mouth to the anus. Anal involvement is rare and is usually accompanied by colitis [47]. Perianal abscess and ulceration have been reported. Treatment involves the full range of topical and systemic anti-inflammatory agents including TNF-alpha inhibitors, calcineurin inhibitors and thalidomide and is sometimes required to be aggressive.

Pyoderma Gangrenosum

This lesion is associated with ulcerative colitis more than Crohn's disease and can be present without either inflammatory bowel disease coexisting. It is rare and presents most commonly on the legs and forearms. Perianal presentation of pyoderma gangrenosum is described but is very rare [48]. It is a purple-edged papular lesion with central ulceration and requires biopsy for diagnosis.

Anal Stenosis

Anal stenosis is a narrowing of the anal canal or the surrounding skin such that there is a reduction in the elasticity or maximum diameter of the orifice. It can be a distressing condition producing difficulties in both evacuation and continence and is challenging to treat.

Aetiology of stenosis of the anal margin is most commonly iatrogenic including haemorrhoidectomy, anal warts excision, radical radiotherapy, congenital anal conditions and Crohn's disease post-surgery (see chapter on Postoperative Anal Conditions). Anal stenosis due to Crohn's is commonly seen within the anal canal rather than at the margin unless postoperative and represents one of the most difficult-to-treat proctological aspects of the disease. The differential diagnosis includes lichen sclerosus, lichen planus and hidradenitis suppurativa and cicatrizing malignant disease.

Inflammatory causes of stricture may be ameliorated with topical anti-inflammatory agents and surgical intervention delayed or prevented. In fibrotic strictures, dilatation may be used. This is performed gently up to 12 mm under anaesthetic if necessary; however, avoidance of sepsis (18 %), sphincter damage and incontinence is a necessarily difficult judgement to make [4]. Surgical management is often necessary, and if the stenosis is superficial and the underlying sphincter mechanism intact, a cutaneous advancement flap may restore elasticity to the perianal skin [49]. If Crohn's disease is present, surgery has a high chance of complications and anal stenosis increases the risk of permanent stoma threefold in Crohn's disease [33].

Skin Pigmentation

Changes in pigmentation alone are not associated with any significant symptoms. Vitiligo is an example which is usually present at other sites when seen perianally. Certain lesions such as AIN and post-inflammatory states can present with hyper or hypopigmentation and biopsy is sometimes required to exclude serious causes. Dermatoses outlined above, most commonly psoriasis can leave pigmentation changes after resolution [50].

Perianal Skin Tags

The process of developing perianal skin tags, elongated areas of skin at anal margin, is usually through thrombosis within the venous complex just deep to the perianal skin in the subcutaneous tissues. At first this produces an exquisitely tender perianal hematoma which in turn stretches the skin, leaving behind an

elongation of normal skin. This is asymptomatic in most, although some patients complain of difficulty cleaning the area, and in some the cosmetic effect disturbs. Highly symptomatic tags should raise concerns regarding underlying Crohn's disease or fistula.

The skin tag so formed is more triangular in shape than the longitudinal sentinel pile seen usually at the posterior anal verge associated with a fissure in ano which is the cause of symptoms, rather than the sentinel pile itself. Due to the sensitive nature of the perianal skin, surgery to remove any skin tags can result in a very painful postoperative course. The tags need to be differentiated from other protuberant lesions of the perianal skin such as condylomata acuminata and more rarely condylomata lata (see later in this chapter).

External Haemorrhoids/ Perianal Hematoma

The acute presentation of a perianal hematoma reveals a blueberry-like lesion at the anal verge which is exquisitely tender. Local anaesthetic application and circumferential incision over the thrombosis with the tip of a blade can lead to evacuation of the thrombosis with often immediate relief of pain and lower recurrence rate [51]. Conservative management is employed for presentation after 48 h of onset.

Radiation Dermatitis

This is usually seen after radiotherapy for anal and low rectal carcinoma but also seen in patients who received radiotherapy for cervical carcinoma in the past. Initial erythema and desquamation results eventually in telangiectasia with effacement and thinning of the skin around the vulva and anal margin. Treatment is symptomatic.

Perianal Ulceration due to Nicorandil

Nicorandil, a potassium channel activator used in the treatment of ischemic heart disease, is a well-recognized rare cause of reversible perianal ulceration [52]. The anal ulcers are usually painful, well defined with undermined edges and refractory to treatment. Awareness of this association may help to avoid unnecessary surgical intervention in patients with angina as liaison with cardiology to substitute the drug for an alternative will relieve the problem.

Rarities

Rarer diseases affecting the perianal region:

1. Baboon syndrome: A distinctive rash in which the initial sensitization is by skin contact with the causative agent, and subsequently the well-defined redness of the buttocks resembling those of baboons occurs on taking the agent by mouth. The patient is generally well and there are no other associated symptoms. Baboon syndrome has been associated with use of nickel and ampicillin. The acronym SDRIFE (symmetrical drug-related intertriginous and flexural erythema) has been used for cases associated with systemic drugs.
2. Pemphigus: Autoimmune chronic bullous disorder which can affect the perianal region usually in the context of widespread bullous changes to the skin.
3. Hailey-Hailey disease (familial benign chronic pemphigus): This autosomal dominant disorder presents commonly in the third or fourth decade. It is characterized by recurrent erosions which become thickened and encrusted and usually involve the flexures. Maceration of skin leads to painful fissures, and secondary bacterial infection can give rise to malodour. Diagnosis can be confirmed with skin biopsy. Treatment includes use of topical corticosteroid creams, oral tetracycline and phototherapy.
4. Darier's disease (keratosis follicularis): An autosomal dominant disorder which presents between 6 and 20 years. Lesions can be skin coloured to red-brown, found usually as scaly papules in seborrhoeic areas. Perianal isolated lesions are unusual but can be large and vegetative with a characteristic malodour. Involvement of the hands is very common. This includes

punctuate keratoses (80 %) and palmar pits (80 %). A V-shaped nick in the nails is a pathognomonic feature. The gene has been identified. Treatment is with topical moisturizers, antiseptics and steroids, ranging to oral retinoids, phototherapy and laser treatments.

5. Acrodermatitis enteropathica: A rare genetic disorder that leads to zinc deficiency and is also seen in adults due to malabsorption of zinc. It presents with red inflamed scaly lesions around body orifices, including the anus, which become crusted and pustular. It is treated with oral zinc.

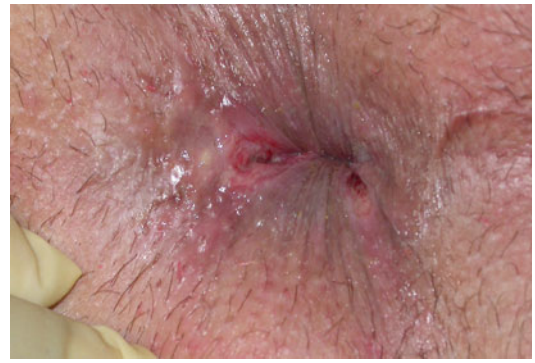

Fig. 20.6 Primary chancre of syphilis. Usually single, but multiple ulcers may be seen. Classically they are indurated and painless (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

Infectious Disorders of Perianal Area

Sexually Transmitted Infections

Infections discussed here are pathogens transmitted primarily through sexual relations, which may be defined as any kind of sexual contact.

Bacterial STIs

Syphilis

Syphilis is caused by *Treponema pallidum* and is an easily missed curable infection causing severe morbidity in a proportion of those untreated. It is also highly transmissible in the early stages, both sexually and vertically from mother to child. Syphilis is common in developing countries and in high-risk groups in developed countries, such as MSM (Men who have sex with men) and sex workers. However, frequent heterosexual outbreaks are reported in recent years outside of these populations [53], meaning all clinicians need to be aware of it.

Syphilis can affect any system of the body. The incubation period is 9–90 days. The lesions seen in the perianal area are:

Primary chancre – usually single, indurated, painless, moist ulcer which develops at the site of inoculation and heals spontaneously (see Fig. 20.6).

Rash of secondary syphilis – erythematous macular and papular lesions, which are often generalized. They are not normally itchy. The palms and soles are frequently involved, but in practice any rash can be syphilis until proven otherwise.

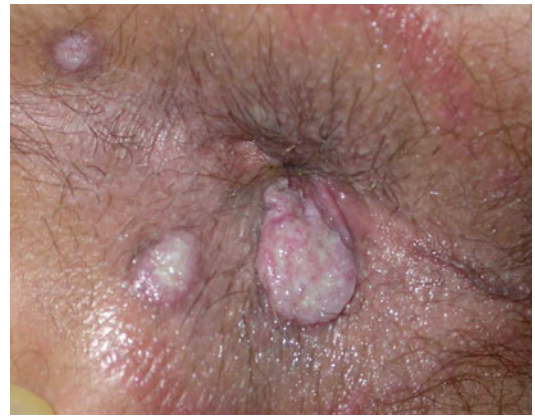

Fig. 20.7 Condyloma lata. Painless, fleshy, soft lesions seen in secondary syphilis (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

Condylomata lata – greyish-white, soft, fleshy, flat-topped moist lesions seen in the genito-anal area, are a feature of secondary syphilis (see Fig. 20.7). These lesions are highly infectious and their exudate is rich in treponemes. If syphilis is suspected, it is important to alert the pathologist of erosive lichen planus to ensure it is not missed on histology.

Serological tests on blood are the simplest way to diagnose syphilis. Specific tests become positive 4–12 weeks after exposure and remain so for life, despite treatment. If very early syphilis infection is suspected, alert the laboratory to perform PCR (polymerase chain reaction) or IgM

tests, which become positive earlier. Dark ground microscopy for direct visualization of treponemes is still used in specialist settings to provide rapid diagnosis of early symptomatic disease.

Intra-muscular benzathine penicillin remains the treatment of choice.

Lymphogranuloma Venereum

Lymphogranuloma venereum (LGV) is caused by the L serovars of *Chlamydia trachomatis*. It has recently become re-established in men who have sex with men (MSM) in developed countries [54]. Reports of perianal and genital ulceration and inguinal buboes as the presenting features in this epidemic are now increasingly emerging [55, 56] although the most common presentation hitherto has been a florid, symptomatic proctitis with systemic features of inflammation. This closely mimics inflammatory bowel disease, and misdiagnoses are reported [57, 58]. Tertiary complications, such as rectal strictures, have been seen where diagnosis has been delayed.

Samples of material from ulcers, buboes or rectal mucosal swabs taken via proctoscopy should be sent for *Chlamydia trachomatis* testing, informing the laboratory that LGV is suspected to ensure that positive samples are referred for specific assays.

Treatment is with doxycycline for a minimum of 3 weeks. Buboes should be drained by aspiration alone. Surgical removal of affected nodes is not helpful.

Other Bacterial STIs

Chlamydia trachomatis and *Neisseria gonorrhoea* remain common STIs affecting the rectum and genital areas but lack perianal skin manifestations.

Chancroid and donovanosis are tropical STIs which should be considered in patients with ulcerative lesions and appropriate travel history. The chancroid ulcer is painful, necrotic with undermined edges and associated with tender inguinal adenopathy [59]. Culture or PCR can be used for diagnosis if available.

Donovanosis causes a chronic, painless, beefy-red, granulomatous ulcer, a differential of a squamous carcinoma [60]. It can be diagnosed on histology with special stains.

Viral STIs

Herpes

Herpes simplex virus (HSV) 2 is the most common cause of genital and perianal ulceration worldwide. The global prevalence is 16 % but with great variation between populations. It is more common in women [61]. An increasing proportion of genital herpes is due to HSV 1, particularly in higher socioeconomic groups where acquisition of perioral HSV 1 in childhood is becoming less common [62]. Both viruses are acquired in the genital region through sexual contact. Latency and recurrence are the hallmark of herpes virus infections, and while there is effective treatment, the virus cannot be eradicated. HSV 2 infection is a major driver of the global human immunodeficiency virus (HIV) epidemic, with infected individuals up to eight times more likely to acquire HIV [63].

HSV classically causes multiple, shallow painful ulcers involving the perianal area alone or extending to the genital area [64]. There may be accompanying proctitis. Primary infection may be severe, lasting 10 days or more and accompanied by systemic symptoms and tender lymphadenopathy. Many individuals with positive HSV serology have never experienced a clinical episode, suggesting that asymptomatic primary infection is common.

Severe HSV infection may be the initial presentation of HIV infection (see Fig. 20.8).

A low threshold for HSV testing is required as lesions are often atypical in appearance [65]. HSV should be considered in the differential diagnosis of perianal skin fissuring as well as ulcerative lesions. A viral swab for HSV PCR (polymerase chain reaction) will confirm the diagnosis and provide typing information. HSV 1 recurs less frequently on the genital region than HSV 2.

Symptomatic HSV can be managed with oral acyclovir alongside analgesics if needed.

Human Papillomaviruses

Human papillomaviruses (HPV) infect the epidermis causing hyperplasia. Over 40 subtypes of HPV have been shown to infect the anogenital skin [66] and are essentially sexually transmitted, although it is theoretically possible that

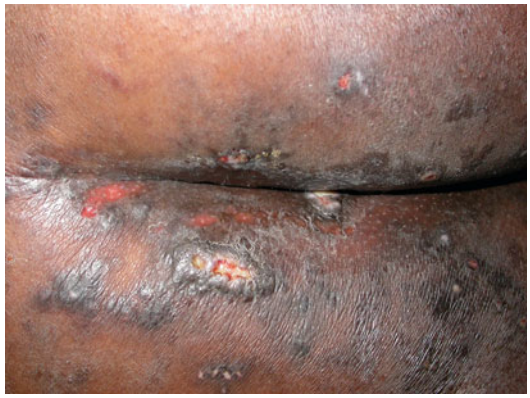

Fig. 20.8 Persistent superficial and necrotic ulcers seen in extensive herpes simplex virus infection (type 2). At presentation, patient also tested positive for HIV (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

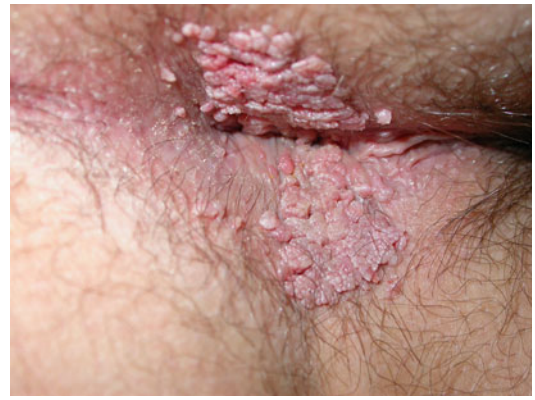

Fig. 20.9 Soft fleshy warts (Courtesy of Dr P N Sashidharan, Homerton University Hospital, London)

transmission from fingers or fomites may occur. Infection occurs via skin-to-skin contact and endures for an average of 2 years before clearance by the immune system [67]. Individuals can be infected with multiple subtypes of HPV, the most common anogenital ones being 6, 11, 16 and 18. Infection may be subclinical or become symptomatic at any time during its course.

The perianal manifestations of anogenital HPV infection include warts and intraepithelial neoplasia (AIN). Anogenital warts are caused by low-risk HPV types (mainly 6 and 11). Warts may be single or multiple, flat or raised and soft or keratotic (See Fig. 20.9). Warts on the perianal skin are common in men and women, including those who have not had anal sexual contact, although they are more common in MSM [68]. In immunosuppressed patients, infection can be prolonged, and therefore, warts may be recurrent over many years. Perianal warts may be associated with irritation and discomfort unlike those at other sites [66]. They do not progress to squamous carcinoma with the possible, very rare exception of giant condyloma acuminata of Buschke and Lowenstein, a locally invasive verrucous carcinoma [69] (see Malignant Disease chapter).

AIN lesions caused by high-risk HPV types (mainly 16, 18) are often asymptomatic but can resemble warts and form part of the differential diagnosis (see Premalignant chapter). Other important differentials are syphilis (condylomata lata), skin tags and Crohn's disease.

Clinical diagnosis is usually sufficient although biopsy can be helpful in atypical cases. Anal smears and anoscopy are under investigation for AIN screening in high-risk subjects [70]. There is no treatment for HPV infection, but locally destructive treatments are used for warts and may reduce infectivity. These can be patient-delivered over a number of weeks, such as podophyllotoxin or imiquimod cream. Cryotherapy may be preferred for warts in the anal canal but requires multiple clinic visits. In particularly large or recalcitrant warts, treatment may include surgical, laser destruction or diathermy resection. Recurrence may occur notwithstanding the method of treatment used.

Vaccines are now available to prevent infection with HPV subtypes 16 and 18, and the quadrivalent vaccine also protects against types 6 and 11. These have been shown to prevent cervical cancer in women [71] and high-grade AIN in MSM [72]. Implementation of the quadrivalent vaccine in Australia led to a significant fall in cases of genital warts in the protected age groups [73].

Molluscum

Molluscum Contagiosum is a pox virus which causes yellowish, dome shaped papules with central umbilication. In adults (but not children) it is usually sexually transmitted and found on genital

skin and upper thighs. Lesions resolve spontaneously after several months but destructive treatments (cryotherapy) can be offered for cosmetic reasons [74]. Extensive lesions may be associated with immunosuppression, particularly HIV.

Human Immunodeficiency Virus (HIV)

HIV infection is the most common cause of immunosuppression worldwide and is eminently treatable. Late diagnosis of HIV remains a significant problem, impacting on mortality and onward transmission. Perianal skin conditions may be the presenting manifestation of HIV. Recalcitrant or severe psoriasis, seborrhoeic dermatitis and recurrent or multi-dermatomal herpes zoster infection should trigger HIV testing, as should AIN or anal carcinoma and Kaposi's sarcoma [17]. Any STI diagnosis should prompt screening for other STIs including HIV.

HIV is associated with aphthous ulceration which can rarely affect the anus, usually with associated oral disease [75].

HIV infection may alter the natural history or appearance of a perianal skin condition. For example, multiple chancres and simultaneous primary and secondary lesions of syphilis can be seen. Herpes simplex lesions may be atypical [76] and recurrences more prolonged and frequent. Anogenital warts may be particularly persistent.

Effective treatment of HIV, restoring immune responses, can paradoxically lead to worsening of existing conditions or precipitate presentation of hitherto occult infections. This is a temporary phenomenon, known as immune reconstitution disease [77]. A common example in the perianal skin is an immune reconstitution reaction to HSV [78], ranging from a short episode to a prolonged, severe ulceration which is resistant to conventional therapy. Other local examples include psoriasis and seborrhoeic dermatitis.

Ectoparasites

Sexually acquired scabies (infection with *Sarcoptes scabiei* mite) presents with intensely pruritic papules, involving the genitalia and often extending to the perineum, buttocks and elsewhere. Infestation with the crab or pubic louse (*Phthirus pubis*) can also lead to severe pruritus in the genital and perianal region with visible lice and eggs. Topical treatment with malathion or permethrin will resolve both infestations, but in scabies itching may persist for a few days after treatment, as it is due to the immune response to mite excrement.

Other Perianal Infections

Bacterial Infections

Pyogenic Bacteria

Staphylococcus aureus and *Streptococcus pyogenes* cause the vast majority of pyogenic skin and soft tissue infections [79] in the perianal region. These infections cause a range of skin and systemic syndromes; those commonly affecting the perianal skin will be described.

Folliculitis is superficial infection of the hair follicles involving the epidermis, which presents with papules, pustules, erosions or crusts and may be itchy. These can coalesce to form a carbuncle, a purulent collection with multiple puncta. Where infection extends to dermal and subcutaneous layers, furuncles are formed. These are clinically palpable, tender nodules with overlying erythema, which suppurate.

Limited folliculitis may respond to topical antibiotics, but extensive disease and furuncles require oral therapy. Abscess requiring surgical management may develop from these conditions.

Cellulitis in the perianal region usually arises following breaches in skin integrity. The infection is usually streptococcal but can be staphylococcal or both. Presentation is with pain, erythema, occasional blistering, oedema and warmth with varying degrees of systemic upset. Oral or intravenous therapy may be needed depending on severity. Isolated streptococcal perianal cellulitis is well described in children, usually lacks systemic features and responds to oral therapy [80].

Sampling for microscopy and culture is recommended to obtain specific organism and resistance information. Empirical therapy with flucloxacillin has traditionally provided cover for

both of these organisms, but the increasing prevalence of MRSA (methicillin-resistant *S. aureus*) limits the utility of this strategy. Local epidemiology and resistance patterns should therefore be consulted when choosing first-line, empirical antibiotics to cover staphylococcal infection. Treatment of choice for *S. pyogenes* infections remains penicillin.

Erythrasma

This intertriginous skin infection is caused by *Corynebacterium minutissimum*, a gram-positive bacillus. It presents as a brownish-red patch affecting the genitocrural area and sometimes the natal cleft. The edge is fairly well defined. The diagnosis is made clinically and can be confirmed with skin scrapings for culture and microscopy but this is seldom necessary. Topical antibiotics such as fusidic acid or clindamycin suffice in limited lesions. More extensive involvement responds rapidly to oral erythromycin [81].

Other Bacterial Infections

Gram-negative infections of the perianal skin are uncommon, but disseminated pseudomonal infection causing ecthyma gangrenosum can occur in sick individuals such as those with leukaemia, particularly children [80].

Mycobacterial skin infection is rare in developed countries but common in settings where tuberculosis (TB) is frequently seen [82]. Perianal skin manifestations are variable and the diagnosis should be considered in unusual or intractable lesions, particularly those with granuloma on histology, and in patients with TB elsewhere or at high risk of TB.

Actinomycosis is a rare bacterial infection [83] causing granulomatous masses or fistulating abscesses perianally, usually in association with pelvic or abdominal disease. Characteristic red or yellow sulphur granules are seen in the pus from these lesions. Penicillin is the treatment of choice.

Viral Infections

Herpes zoster, due to varicella-zoster virus, is an important differential diagnosis of ulceration in the perianal region. The vesicular lesions are usually unilateral in a dermatomal distribution, with S3 affecting the buttock and S4 and 5 affecting the perianal skin. Diagnosis can be confirmed with viral culture or PCR. Treatment with high-dose oral acyclovir or alternatives is effective and reduces the risk of post-herpetic neuralgia [84], a common complication.

Cytomegalovirus can also cause painful ulceration in immunosuppressed individuals and is diagnosed on histology. It is treated with ganciclovir.

Fungal Infections

Candida species, usually *Candida albicans*, are a common cause of intertrigo which can involve the perianal area. This presents with erythema in moist skin folds, associated with characteristic satellite lesions and may coexist with other causes of intertrigo. Treatment is with topical or systemic imidazoles, depending on the extent of disease. Cultures should be performed in recalcitrant disease to identify alternative species such as *C. glabrata*, which may be imidazole resistant.

Tinea cruris is usually caused by dermatophytes: *Trichophyton*, *Epidermophyton* or *Microsporum* species. These infections produce a well-defined patch with raised erythematous borders, beginning in the groins and extending to the buttocks and perianally. The rash is itchy and spreads outwards with partial central clearing. Skin scrapings for microscopy and culture can confirm the diagnosis. Topical imidazoles are usually sufficient treatment.

Perianal ulceration has been reported with a number of systemic mycoses such as *Histoplasma capsulatum* [85] and *Blastomyces dermatitidis* [86]. Fungi are important to consider in a debilitated patient with ulceration or umbilicated papular lesions. Fungal stains and cultures on biopsies will identify these diseases.

Protozoal Infections

Entamoeba histolytica is transmitted oro-faecally and usually causes colitis but can also cause amoebiasis cutis, a granulomatous ulcer or abscess usually in the perianal region [87]. Diagnosis is via microscopic demonstration of

trophozoites in material from the lesion. Metronidazole is the standard treatment.

Helminth Infections

Strongyloides stercoralis is a common chronic tropical infection which may be seen in migrants or travellers [88]. The larvae migrate under the skin and produce a characteristic moving rash, common perianally and on the buttocks, called larva currens. Serology will confirm the diagnosis.

Schistosomiasis is a tropical worm infection acquired through contact with contaminated fresh water [89]. Perianal lesions result from ectopic egg deposition in skin and are granulomatous. They can be papular, resembling warts, or ulcerated. It is also a rare differential of fistula-in-ano [90]. Diagnosis can be made on biopsy.

Conclusion

Perianal skin disorders provide challenges to the proctologist through their wide range of diverse causes. Several systemic skin conditions such as psoriasis can present in this area, making examination of extra-anal sites prudent, particularly the genital region. Sexually transmitted infections and HIV may also present perianally and must be diagnosed and treated in a timely fashion to prevent onward transmission and, in the case of HIV, avoidable mortality. HIV testing should be offered to all with unusual or recalcitrant lesions or who are known to be at risk. Liaison with other specialties such as dermatology or sexual health may be needed to optimize the management of patients with perianal skin disease.

References

1. Phillips R, Brown T. Surgical management of anal incontinence Part A: secondary anal sphincter repair. In: Sultan A, Thakar A, Fenner D, editors. Perineal and anal sphincter trauma. London: Springer; 2007. p. 144–53.
2. Dodi G, Pirone E, Bettin A, et al. The mycotic flora in proctological patients with and without pruritus ani. Br J Surg. 1985;72(12):967–9.
3. Schug S, Raajkumar A. Local anesthetics. In: Aronson J, editor. Meyler's side effects of drugs used in anesthesia. Amsterdam: Elsevier; 2008. p. 152–63.
4. Bouguen G, Siproudhis L, Bretagne JF, Bigard MA, Peyrin-Biroulet L. Nonfistulizing perianal Crohn's disease: clinical features, epidemiology, and treatment. Inflamm Bowel Dis. 2010;16(8):1431–42.
5. Grant A, Gonzalez T, Montgomery MO, Cardenas V, Kerdel FA. Infliximab therapy for patients with moderate to severe hidradenitis suppurativa: a randomized, double-blind, placebo-controlled crossover trial. J Am Acad Dermatol. 2010;62(2):205–17.
6. Herbenick D, Reece M, Schick V, Sanders SA, Dodge B, Fortenberry JD. Sexual behavior in the United States: results from a national probability sample of men and women ages 14–94. J Sex Med. 2010;7 Suppl 5:255–65.
7. Johnson AM, Mercer CH, Erens B, et al. Sexual behaviour in Britain: partnerships, practices, and HIV risk behaviours. Lancet. 2001;358(9296):1835–42.
8. Fornasa CV, Trento A, Bezze G. Pruritus ani: etiologic and causative factors in 94 adult patients. Pelviperineology. 2007;26(4):178–9.
9. Schnuch A, Carlsen B. Genetics and individual predispositions. In: Johansen J, Frosch P, Lepoittevin J, editors. Contact dermatitis. New York: Springer; 2010. p. 13–42.
10. Veien N. Clinical features. In: Johansen J, Frosch P, Lepoittevin J, editors. Contact dermatitis. New York: Springer; 2010. p. 255–303.
11. Bauer A, Geier J, Elsner P. Allergic contact dermatitis in patients with anogenital complaints. J Reprod Med. 2000;45(8):649–54.
12. Kugler K, Brinkmeier T, Frosch PJ, Uter W. Anogenital dermatoses–allergic and irritative causative factors. Analysis of IVDK data and review of the literature. J Dtsch Dermatol Ges. 2005;3(12):979–86.
13. Lazzarini R, Duarte I, de Farias DC, Santos CA, Tsai AI. Frequency and main sites of allergic contact dermatitis caused by nail varnish. Dermatitis. 2008;19(6):319–22.
14. Gardner KH, Davis MD, Richardson DM, Pittelkow MR. The hazards of moist toilet paper: allergy to the preservative methylchloroisothiazolinone/methylisothiazolinone. Arch Dermatol. 2010;146(8):886–90.
15. Vermaat H, Smienk F, Rustemeyer T, Bruynzeel DP, Kirtschig G. Anogenital allergic contact dermatitis, the role of spices and flavour allergy. Contact Dermatitis. 2008;59(4):233–7.
16. Warshaw EM, Furda LM, Maibach HI, et al. Anogenital dermatitis in patients referred for patch testing: retrospective analysis of cross-sectional data from the North American Contact Dermatitis Group, 1994–2004. Arch Dermatol. 2008;144(6):749–55.
17. British HIV Association, British Association of Sexual Health and HIV, British Infection Society. UK national guidelines for HIV testing, 2008. London: BHIVA/BASHH/BIS; 2008.

18. Virgili A, Bacilieri S, Corazza M. Evaluation of contact sensitization in vulvar lichen simplex chronicus. J Reprod Med 2003:48:33–6.
19. Crone AM, Stewart EJ, Wojnarowska F, Powell SM. Aetiological factors in vulvar dermatitis. J Eur Acad DermatolVenereol 2000:14:181–6.
20. Oyama N, Chan I, Neill SM, et al. Autoantibodies to extracellular matrix protein 1 in lichen sclerosus. Lancet. 2003;362(9378):118–23.
21. Cheng S, Kirtschig G, Cooper S, Thornhill M, Leonardi-Bee J, Murphy R. Interventions for erosive lichen planus affecting mucosal sites. Cochrane Database Syst Rev. 2012;(2):CD008092.
22. Abstracts from "Directions 2006: The First International Hidradenitis Suppurativa Research Symposium". Exp Dermatol. 2006;15:478. doi: 10.1111/j.0906-6705.2006.0436a.x
23. Von Der Werth JM, Williams HC, Raeburn JA. The clinical genetics of hidradenitis suppurativa revisited. Br J Dermatol. 2000;142(5):947–53.
24. Plewig G, Kligman A. Acne: morphogenesis and treatment. Berlin: Springer; 1975.
25. Stellon AJ, Wakeling M. Hidradenitis suppurativa associated with use of oral contraceptives. BMJ. 1989;298(6665):28–9.
26. Hurley HJ. Axillary hyperhidrosis, apocrine bromhidrosis, hidradenitis suppurativa, and familial benign pemphigus: surgical approach. In: Roenigk RK, Roenigk HH, editors. Dermatologic surgery. New York: Marcel Dekker; 1989.
27. Mortimer PS, Lunniss PJ. Hidradenitis suppurativa. J R Soc Med. 2000;93(8):420–2.
28. Brocard A, Knol AC, Khammari A, Dreno B. Hidradenitis suppurativa and zinc: a new therapeutic approach. A pilot study. Dermatology. 2007;214(4):325–7.
29. Boer J, Jemec GB. Resorcinol peels as a possible self-treatment of painful nodules in hidradenitis suppurativa. Clin Exp Dermatol. 2010;35(1):36–40.
30. Rambhatla PV, Lim HW, Hamzavi I. A systematic review of treatments for hidradenitis suppurativa. Arch Dermatol. 2012;148(4):439–46. doi: 10.1001/archdermatol.2011.1950. Epub 2011 Dec 19.
31. van Rappard DC, Limpens J, Mekkes JR. The off-label treatment of severe hidradenitis suppurativa with TNF-alpha inhibitors: a systematic review. J Dermatolog Treat. 2013;24(5):392–404. doi: 10.3109/09546634.2012.674193. Epub 2012 Apr 12.
32. Eglinton TW, Roberts R, Pearson J, Barclay M, Merriman TR, Frizelle FA, Gearry RB. Clinical and genetic risk factors for perianal Crohn's disease in a population-based cohort. Am J Gastroenterol. 2011. doi:10.1038/ajg.2011.437.
33. Galandiuk S, Kimberling J, Al-Mishlab TG, Stromberg AJ. Perianal Crohn disease: predictors of need for permanent diversion. Ann Surg. 2005; 241(5):796–801; discussion 801–2.
34. Peyrin-Biroulet L, Loftus Jr EV, Tremaine WJ, Harmsen WS, Zinsmeister AR, Sandborn WJ. Perianal Crohn's disease findings other than fistulas in a population-based cohort. Inflamm Bowel Dis. 2012; 18(1):43–8.
35. Sandborn WJ, Fazio VW, Feagan BG, Hanauer SB. AGA technical review on perianal Crohn's disease. Gastroenterology. 2003;125(5):1508–30.
36. Peyrin-Biroulet L, Loftus Jr EV, Tremaine WJ, Harmsen WS, Zinsmeister AR, Sandborn WJ. Perianal Crohn's disease findings other than fistulas in a population-based cohort. Inflamm Bowel Dis. 2011;18(1):43–8.
37. Hughes LE. Surgical pathology and management of anorectal Crohn's disease. J R Soc Med. 1978;71(9):644–51.
38. Irvine EJ. Usual therapy improves perianal Crohn's disease as measured by a new disease activity index. McMaster IBD Study Group. J Clin Gastroenterol. 1995;20(1):27–32.
39. Mahadev S, Young JM, Selby W, Solomon MJ. Self-reported depressive symptoms and suicidal feelings in perianal Crohn's disease. Colorectal Dis. 2012;14(3): 331–5.
40. Ingle SB, Loftus Jr EV. The natural history of perianal Crohn's disease. Dig Liver Dis. 2007;39(10): 963–9.
41. Williams DR, Coller JA, Corman ML, Nugent FW, Veidenheimer MC. Anal complications in Crohn's disease. Dis Colon Rectum. 1981;24(1):22–4.
42. Fleshner PR, Schoetz Jr DJ, Roberts PL, Murray JJ, Coller JA, Veidenheimer MC. Anal fissure in Crohn's disease: a plea for aggressive management. Dis Colon Rectum. 1995;38(11):1137–43.
43. Lozynskyy YS. Treatment algorithms in the case of perianal complications of Crohn's disease. Dig Dis. 2009;27(4):565–70.
44. Siproudhis L, Mortaji A, Mary JY, Juguet F, Bretagne JF, Gosselin M. Anal lesions: any significant prognosis in Crohn's disease? Eur J Gastroenterol Hepatol. 1997;9(3):239–43.
45. Nivatvongs S, Gordon P. Crohn's Disease. In: Nivatvongs S, Gordon P, editors. Principles and practice of surgery for the colon, rectum, and anus. 3rd ed. New York: Informa Healthcare; 2007. p. 819–908.
46. Church JM, Fazio VW, Lavery IC, Oakley JR, Milsom JW. The differential diagnosis and comorbidity of hidradenitis suppurativa and perianal Crohn's disease. Int J Colorectal Dis. 1993;8(3):117–9.
47. Lee RG. The colitis of Behcet's syndrome. Am J Surg Pathol. 1986;10(12):888–93.
48. Burgess NA, Lari JL. Pyoderma gangrenosum with large circumferential perianal skin loss in a child. Br J Clin Pract. 1991;45(3):223–4.
49. Habr-Gama A, Sobrado CW, de Araujo SE, et al. Surgical treatment of anal stenosis: assessment of 77 anoplasties. Clinics (Sao Paulo). 2005;60(1): 17–20.
50. Stein E. Perianal pigmentary disorders. In: Stein E. Anorectal and Colon Diseases. Berlin, Heidelberg: Springer-Verlag; 2003. pp 121–7.
51. Greenspon J, Williams SB, Young HA, Orkin BA. Thrombosed external hemorrhoids: outcome after conservative or surgical management. Dis Colon Rectum. 2004;47(9):1493–8.

52. Akbar F, Maw A, Bhowmick A. Anal ulceration induced by nicorandil. BMJ. 2007;335(7626): 936–7.
53. Simms I, Bell G, Hughes G. Infectious syphilis in young heterosexuals: responding to an evolving epidemic. Int J STD AIDS. 2011;22(9):481–2.
54. White JA. Manifestations and management of lymphogranuloma venereum. Curr Opin Infect Dis. 2009; 22(1):57–66.
55. Singhrao T, Higham E, French P. Lymphogranuloma venereum presenting as perianal ulceration: an emerging clinical presentation? Sex Transm Infect. 2011;87(2):123–4.
56. Sethi G, Allason-Jones E, Richens J, et al. Lymphogranuloma venereum presenting as genital ulceration and inguinal syndrome in men who have sex with men in London, UK. Sex Transm Infect. 2009;85(3):165–70.
57. Soni S, Srirajaskanthan R, Lucas SB, Alexander S, Wong T, White JA. Lymphogranuloma venereum proctitis masquerading as inflammatory bowel disease in 12 homosexual men. Aliment Pharmacol Ther. 2010;32(1):59–65.
58. Hoie S, Knudsen LS, Gerstoft J. Lymphogranuloma venereum proctitis: a differential diagnose to inflammatory bowel disease. Scand J Gastroenterol. 2010;46(4):503–10.
59. Lewis DA. Chancroid: clinical manifestations, diagnosis, and management. Sex Transm Infect. 2003; 79(1):68–71.
60. O'Farrell N, Moi H, IUSTI/WHO European STD guidelines Editorial Board. European guideline for the management of donovanosis, 2010. Int J STD AIDS. 2010;21(9):609–10.
61. Looker KJ, Garnett GP, Schmid GP. An estimate of the global prevalence and incidence of herpes simplex virus type 2 infection. Bull World Health Organ. 2008;86(10):805–12, A.
62. Malkin JE. Epidemiology of genital herpes simplex virus infection in developed countries. Herpes. 2004;11 Suppl 1:2A–2323.
63. Ward H, Ronn M. Contribution of sexually transmitted infections to the sexual transmission of HIV. Curr Opin HIV AIDS. 2010;5(4):305–10.
64. Sissons JGP. Oxford Textbook of Medicine. Chapter 7.5.2 Herpesviruses (excluding Epstein Barr virus). In: Warrell DA, Cox TM, Firth JD, editors. 5th ed. Oxford, New York: Oxford University Press; 2011. Accessed 9/2/2012.
65. Tyring SK, Carlton SS, Evans T. Herpes. Atypical clinical manifestations. Dermatol Clin. 1998;16(4): 783–8, xiii.
66. Winer R, Koutsky LA. Genital human papillomavirus infections. In: Holmes KK, Sparling P, Stamm W, editors. Sexually transmitted infections. 4th ed. New York: McGraw-Hill; 2007.
67. Pattman R. Oxford handbook of genitourinary medicine, HIV, and sexual health. 2nd ed. Oxford ; New York: Oxford University Press; 2010.
68. Sonnex C, Scholefield JH, Kocjan G, et al. Anal human papillomavirus infection in heterosexuals with genital warts: prevalence and relation with sexual behaviour. BMJ. 1991;303(6812):1243.
69. Martin JM, Molina I, Monteagudo C, Marti N, Lopez V, Jorda E. Buschke-Lowenstein tumor. J Dermatol Case Rep. 2008;2(4):60–2.
70. Oon SF, Winter DC. Perianal condylomas, anal squamous intraepithelial neoplasms and screening: a review of the literature. J Med Screen. 2010; 17(1):44–9.
71. Future I II Study Group, Dillner J, Kjaer SK, et al. Four year efficacy of prophylactic human papillomavirus quadrivalent vaccine against low grade cervical, vulvar, and vaginal intraepithelial neoplasia and anogenital warts: randomised controlled trial. BMJ. 2010;341:c3493.
72. Palefsky JM, Giuliano AR, Goldstone S, et al. HPV vaccine against anal HPV infection and anal intraepithelial neoplasia. N Engl J Med. 2011;365(17): 1576–85.
73. Read TR, Hocking JS, Chen MY, Donovan B, Bradshaw CS, Fairley CK. The near disappearance of genital warts in young women 4 years after commencing a national human papillomavirus (HPV) vaccination programme. Sex Transm Infect. 2011;87(7):544–7.
74. Brown M, Paulson C, Henry SL. Treatment for anogenital molluscum contagiosum. Am Fam Physician. 2009;80(8):864.
75. Kerr AR, Ship JA. Management strategies for HIV-associated aphthous stomatitis. Am J Clin Dermatol. 2003;4(10):669–80.
76. Ranu H, Lee J, Chio M, Sen P. Tumour-like presentations of anogenital herpes simplex in HIV-positive patients. Int J STD AIDS. 2011;22(4):181–6.
77. Lawn SD, Meintjes G. Pathogenesis and prevention of immune reconstitution disease during antiretroviral therapy. Expert Rev Anti Infect Ther. 2011;9(4): 415–30.
78. Couppie P, Sarazin F, Clyti E, et al. Increased incidence of genital herpes after HAART initiation: a frequent presentation of immune reconstitution inflammatory syndrome (IRIS) in HIV-infected patients. AIDS Patient Care STDS. 2006;20(3): 143–5.
79. Tognetti L, Martinelli C, Berti S, et al. Bacterial skin and soft tissue infections: review of the epidemiology, microbiology, aetiopathogenesis and treatment: A collaboration between dermatologists and infectivologists. J Eur Acad Dermatol Venereol. 2012;26:931–41.
80. Hay R. Oxford Textbook of Medicine. Chapter 23.10 Infections and the skin. In: Warrell DA, Cox TM, Firth JD, editors. 5th ed. Oxford: Oxford University Press; 2011. Accessed 9/2/2012.
81. Holdiness MR. Management of cutaneous erythrasma. Drugs. 2002;62(8):1131–41.
82. Bravo FG, Gotuzzo E. Cutaneous tuberculosis. Clin Dermatol. 2007;25(2):173–80.
83. Wong VK, Turmezei TD, Weston VC. Actinomycosis. BMJ. 2011;343:d6099.
84. Schmader KE, Dworkin RH. Natural history and treatment of herpes zoster. J Pain. 2008;9(1 Suppl 1):S3–9.

85. Recondo G, Sella A, Ro JY, Dexeus FH, Amato R, Kilbourn R. Perianal ulcer in disseminated histoplasmosis. South Med J. 1991;84(7):931–2.
86. Ricciardi R, Alavi K, Filice GA, Heilly SD, Spencer MP. Blastomyces dermatitidis of the perianal skin: report of a case. Dis Colon Rectum. 2007;50(1):118–21.
87. Stanley Jr SL. Amoebiasis. Lancet. 2003;361(9362):1025–34.
88. Ganesh S, Cruz Jr RJ. Strongyloidiasis: a multifaceted disease. Gastroenterol Hepatol (N Y). 2011;7(3):194–6.
89. Gryseels B, Polman K, Clerinx J, Kestens L. Human schistosomiasis. Lancet. 2006;368(9541):1106–18.
90. Georghiou PR, Tilse MH, McCormack JG, Neely MG. Fistula-in-ano: an unusual presentation of chronic schistosomiasis. Aust N Z J Surg. 1995;65(7):541–3.

Pruritus Ani

21

Tim Brown

Happiness is having a scratch for every itch.
Ogden Nash
American 20th Century Poet
Died 1971 of complications related to Crohn's Disease.

Introduction

Pruritus ani, in its worst form, is a miserable affliction. It is common; however, there is a spectrum of symptom severity and those falling into the severe category are rare. The majority of patients experience nothing more troubling than a transient 'itchy bottom'. Simple measures are usually all that are required to relieve suffering, and their physician is not consulted. Other patients with more persistent symptoms present to the proctologist who is confronted then with a difficult problem and a long list of differential diagnoses to consider and investigate.

Biology of Itch

Itch can be defined as an unpleasant cutaneous sensation associated with an urge to scratch. It is more or less voluntary, yet can very often be a subconscious motor activity [1].

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The sensation of itch may have evolved in order to protect the skin from agents (e.g. parasites, plant toxins) that would potentially breach its barrier to harm the organism. In this way, the itch response has similarities to the pain response, and indeed itch (pruriceptive) and pain (nociceptive) pathways seem to have evolved in tandem.

However, the two have differences. Whereas the sensation of pain causes reflex withdrawal away from the source of pain in an attempt to avoid the agent, itching causes an opposite response, that being to scratch, perhaps in an attempt to rid the skin of an agent that has already breached the skin's defences.

Pruriceptive and nociceptive pathways have evolved in order to provide us with potential survival benefits. As with other body systems that have evolved over generations, the pruriceptive system can become maladaptive in a number of individuals. This is usually in response to one or more causative agents, manifesting as miserable, intractable itching [1].

Itch is generated by specialised, itch-dedicated, cutaneous unmyelinated C fibres that have dense sensory nerve endings. These are distinct from the polymodal nociceptor mechanoreceptors involved in pain signalling as they do not respond to heat, mechanical or chemical stimuli. They show a sustained response to histamine which is a potent pruritogen and express the cell membrane receptor transient receptor potential cation channel subfamily V member 1 (TrpV1), also known as the capsaicin receptor [2].

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Pruriceptive fibres ascend the spinothalamic tract into the thalamus which in turn has projections into the cerebral cortex. Spinal and higher cortical projections interact with ascending fibres, and it is hypothesised that these projections attenuate the itch signal, much like the gating mechanism that exists for pain perception. Ikoma et al. have written an excellent and comprehensive review on the neurobiology of itch [3].

On a behavioural level, patients undergo a vicious itch-scratch-itch cycle. A pruritogen causes the skin to itch. The itch drives a scratch reflex. Scratching traumatises the skin and induces pain. Pain in turn suppresses itch for a length of time but the skin damage stimulates the release of pruritogenic inflammatory mediators leading to further itching and scratching and so on ad infinitum. The desire to scratch can be denied as there is a degree of higher cortical control over motor function; however, a stronger and stronger desire to satisfy the urge to scratch develops until the sufferer must relent. What follows is a vigorous and traumatising episode of scratching that produces a feeling that can be described as a combination of guilt, pain and exquisite pleasure. These symptoms are not confined to daylight hours. Patients often wake from sleep to find their fingernails bleeding, having traumatised their skin by scratching vigorously during sleep.

Pruritus Ani

Pruritus may be a reflection of an underlying systemic disease, a primary dermatological illness, a psychiatric or behavioural problem or a condition affecting the anorectum [4]. These causes and their various management options are discussed in detail elsewhere in this book but are summarised in Box 21.1. If investigation reveals no specific condition to be causing their symptoms, the patients are diagnosed as having idiopathic pruritus ani, a notoriously difficult and depressing condition to contend with, both as a patient and as that patient's physician. This chapter will deal specifically with idiopathic pruritus ani (IPA).

Box 21.1 Infective

Bacterial

Staphylococcus aureus, beta haemolytic streptococcus, *Corynebacterium minutissimum*, lymphogranuloma venereum, syphilis, tuberculosis, actinomycosis

Viral

Herpes simplex, herpes zoster, cytomegalovirus, human immunodeficiency virus, molluscum contagiosum, condylomata acuminata (papillomavirus)

Fungal

Candida albicans

Parasitic

Enterobius vermicularis (oxyuriasis, pinworm), Schistosomiasis cutis, *Sarcoptes scabiei* (scabies)

Neoplastic

Squamous cell carcinoma, basal cell carcinoma, Bowen's disease, extra mammary Paget's disease, melanoma, mycosis fungoides

Dermatoses

Psoriasis, lichen planus, seborrhoeic dermatitis, atopic dermatitis, erythema multiforme, systemic lupus erythematosus, amyloidosis, radiation dermatitis, lichen sclerosus et atrophicus, contact dermatitis, allergic dermatitis, scleroderma

Contact irritant

Drugs (e.g. IV steroid), topical applications, soap and cosmetics, clothing, detergents, latex

Anorectal

Fistula in ano, diarrhoeal illness, fissure in ano, haemorrhoids, gutter deformity, primary or secondary sphincter dysfunction, fibroepithelial polyp, villous adenoma, rectal or anal malignancy, postanal canal surgery

Systemic disease

Liver disease, renal failure, polycythemia rubra vera, diabetes mellitus, leukaemia

Psychological

Depression, psychosomatic illness, obsessive-compulsive disorder

Idiopathic Pruritus Ani

The true incidence of IPA is difficult to establish but in general is considered to be common albeit with a wide spectrum of severity. Men are afflicted more than women in a ratio of 4:1 [5].

A number of theories have been postulated in an attempt to describe an aetiological cause for pruritic symptoms.

Dietary

Specific dietary factors have been reported as important as a causative factor that once removed sees resolution of symptoms [6]. There is, however, little robust evidence for this and reports are largely anecdotal. Caffeine in particular has been reported as an irritant as well as being reported to cause transient weakness in the anal sphincter after its ingestion [7, 8]. In this, there may be an explanation for pruritic symptoms in the setting of subclinical incontinence (see next section).

Faecal Contamination

Poor perianal hygiene has been implicated as a cause of pruritus ani [7]. In an elegant experiment, Caplan applied autologous faeces to the perianal and underarm skin of a group of patients with ($n=12$) and a group of patients without ($n=15$) pruritus ani. A further group ($n=10$) had topical faecal application simulated to act as control [9].

Twelve of 27 of these subjects complained of perianal itching with an onset between 1 and 6 h of faecal application to the perianal skin. Four of these subjects had a history of pruritus ani, eight had no prior history. None of the control group suffered symptoms. Pruritus was instantly relieved with cleansing. A single subject developed pruritus on application to the arm. The conclusion of the study was that faeces acted as an irritant rather than an allergen.

Farouk studied rectal and internal anal sphincter pressures in a group of pruritus ani patients [10]. Those with pruritus had higher rectal pressures with lower internal anal sphincter pressures and prolonged internal sphincter relaxation than the control group. Pruritus was reported within an hour of the abnormal internal sphincter relaxation. The authors' conclusion was that occult faecal leakage was a cause of pruritus secondary to abnormal internal sphincter relaxation.

Given the above findings, chronic leakage of irritant faeces causing itch with subsequent mechanical skin trauma makes an interesting hypothesis. If indeed this is the case, symptoms may be amenable to treatment with a stool thickener such as loperamide. No trial data have been presented to support the hypothesis although anecdotally this approach can be successful.

Infective

The perianal region is subject to the same skin commensals as the remainder of the body. Due to its anatomy within the warm, moist gluteal folds and at the outlet from the gastrointestinal tract, additional flora may exist and thrive. Bacterial, viral, fungal and parasitic organisms all have been implicated as an aetiology; therefore, thorough investigation with swab, scraping and Wood's light examination is essential. Sexually transmitted infection is common and therefore appropriate questioning on history taking is essential.

Contact Dermatitis and Occult Perianal Dermatology

Dasan reports an interesting series of consecutive patients presenting to a combined dermatological and coloproctological clinic [11]. Out of 40 patients, 2 were identified as suffering from an anorectal condition that required surgical intervention. Thirty-four out of 40 patients were suffering from an underlying dermatosis, treatment of which improved or resolved their symptoms. Patch testing was undertaken in 32 out of 40. Eighteen of these patients showed hypersensitivity to allergens which are commonly found in remedies for pruritus ani. A patient was found to be sensitive to an ingredient of his wife's shampoo. Symptoms resolved on cessation of her practice of washing her hair in their shared bath water.

Patch testing as a useful instrument in the investigation of chronic pruritus ani is supported by Harrington who tested 80 patients with PA [12]. Fifty-five of these patients were patch test 'positive', 38 of them for a medication commonly used as a remedy for pruritus. As well as topical preparations, the advice to use 'wet wipes' is commonly given to patients in the clinic. Ingredients of wet wipes are occasionally allergenic on patch testing [13] and should be avoided.

Underlying Proctological Disease

Daniel et al. report in their series of 109 patients with pruritus ani that 75 % of these had an underlying coloproctological disorder: 20 % had haemorrhoids and 12 % had anal fissures; however, 19 % had an underlying coloproctological malignancy (11 % rectal cancer, 6 % anal cancer, 2 % colonic cancer) [7]. It is interesting to note this group's definition of chronic pruritus ani as being a condition with symptoms lasting over 6 weeks. Mentes' group had a median symptom length of 24 months [14]. Underlying anorectal conditions, whilst clearly important to exclude and treat if appropriate, do not seem to be as prevalent in other groups investigating idiopathic pruritus ani with a longer time course [11, 14–16].

Approach to Idiopathic Pruritus Ani

Having gone through appropriate examination, diagnosis and management of any identifiable cause of pruritus, a proportion of patients will remain symptomatic. These patients are defined as having idiopathic pruritus ani and can be challenging to manage.

Goligher's feelings towards pruritic patients were highlighted in Sagar's paper [15]:

> …a rectal clinic is apt to be haunted by its pruritic patients.. the peri-anal skin may be painful even to look at.. the itch has a tormenting, distracting character.. pain by comparison is almost a pleasure.. a bizarre form of auto-eroticism may result.. it is difficult to be enthusiastic about its treatment.

The clinician is hampered by a lack of knowledge regarding the underlying aetiology of this affliction and lacks a universally acceptable and easily deployed treatment that will alleviate the relentless suffering experienced by these patients. When faced with a pruritic patient in the clinic, therefore, the frustration of seeing such an individual is understandable.

However, some treatments that are generating interest and displaying promise have come to light since Goligher issued his statement.

Topical Capsaicin

Capsaicin is a biochemical extracted from red chilli peppers that has found success in the management of chronic pain. It is the active ingredient that puts the 'heat' into curries or other spicy foods. Its mechanism of action, although not completely understood, suggests that it plays a role in reducing substance 'p' concentrations from presynaptic neurones. Substance 'p' is an important sensory neuropeptide that may be responsible for transmission of signals along 'itch'-specific, type 'C' sensory neurones [17].

At its standard topical dose of 0.025 %, capsaicin causes an intense burning sensation when applied topically. This sensation prevents its use on perianal skin as the pain is poorly tolerated. Lysy's group felt that capsaicin may still have a role to play in the treatment of pruritus ani, albeit at an attenuated dose. By diluting the concentration, they were able to demonstrate that it was in fact tolerable to most patients and, when compared to a menthol ointment preparation, was effective as a treatment [18].

In their study, Lysy et al. studied 44 patients who had been diagnosed as having idiopathic pruritus ani. These patients underwent a double-blind, placebo-controlled crossover trial of capsaicin (0.006 %) ointment versus a menthol ointment as a placebo, each applied three times daily. Each arm of the trial lasted 4 weeks. Outcomes were measured on the basis of a symptom diary.

Results of the trial report that 31/44 patients experienced relief of itching with capsaicin versus 0/44 with placebo ($p < 0.0001$). For 24 of

these patients, relief was achieved within 24 h of commencing treatment, the remaining 7 experiencing relief within 72 h.

Long-term follow-up was achieved for 18 patients. Remission from symptoms was achieved with regular application every 2–3 days. Relief was either complete (4/18) or almost complete (14/18). Loss of effect was experienced in two patients who responded to increasing the concentration of the topical ointment to 0.012 %.

All patients experienced a 'burning' sensation as a side effect. Four patients were so intolerant of this symptom that they were unable to complete the trial. One patient developed urticaria and also was excluded. The burning sensation diminished with prolonged application.

Intradermal Methylene Blue Injection (Anal Tattooing Procedure) as a Treatment for Idiopathic Pruritus Ani

Methylene blue was the first synthetic drug and has found a variety of uses in medicine and industry over the last 120 years. Schirmer et al. present an excellent summary of the history, biochemical properties and various clinical applications of methylene blue [19].

In 1973, Yaacov Wolloch made a chance discovery of a technique involving local subcutaneous injection of methylene blue as a treatment for pruritus ani. The source was Rygick's 'Atlas of the operations on the rectum and colon' which was published in the former USSR. As the technique appeared to be straightforward, Wolloch and Dintsman adopted it and reported complete success in the treatment of 8 of 9 patients suffering from IPA in 1979 [20]. Their method involved outpatient treatment under local injection of 2 % procaine infiltration followed by the subcutaneous infiltration of 15–20 ml of 15 % methylene blue solution. The only reported side effect was a transient pyrexia in a single patient.

The technique continued to be unreported in the literature until Eusebio, Graham and Mody reported their experience between 1979 and 1989 on 21 patients with IPA. After 9.5 years of follow-up, recurrence was noted in 4 patients [21].

Initially, a mixture of 30–40 ml of 0.25 % Marcaine with 1:200,000 epinephrine was infiltrated, followed by 30 ml of 0.5 % methylene blue when topical anaesthesia had been achieved. Cellulitis was observed in four of the patients and full-thickness necrosis requiring formal debridement was observed in three patients.

To address the complications, the authors modified their technique by injecting a mixture of 10 ml 1 % methylene blue, 5 ml normal saline, 7.5 ml of 0.25 % Marcaine with 1:200,000 epinephrine and 7.5 ml of 0.5 % Xylocaine under intravenous sedation. There were no further incidences of cellulitis or skin necrosis [22].

The authors observed that loss of pin-prick sensation in the perianal area was predominant amongst the treated patients. A punch biopsy of the perianal skin at 7 years of follow-up demonstrated normal nerve axons but no sensory nerve endings on electron microscopy. Toxicity of methylene blue to nerve endings has been hypothesised as the therapeutic mode of action.

Farouk and Lee report a small series of six consecutive patients who underwent injection of methylene blue [23]. A solution of 10 ml of methylene blue (1 %) was mixed with 7.5 ml Marcaine (0.25 %) with adrenaline (1:200,000) and 0.5 % Marcaine plus 5 ml 0.9 % saline solution. This mixture was injected intradermally under general anaesthesia with prophylactic antibiotic cover (cefuroxime, 750 mg, and metronidazole, 500 mg).

Five of the six patients experienced a substantial reduction in symptoms after the injection. Patients remained under long-term follow-up and each received subsequent injections up to 5 years after initial treatment.

All patients reported numbness in the treated skin following the procedure which was tolerable.

Botterill and Sagar report a series of 25 patients with intractable pruritus ani [15]. Most (23/25) of these patients had undergone previous procedures for anorectal pathology believed to be contributing to their pruritus. The authors used a mixture of 15 ml 1 % lidocaine, 5 ml 1 % methylene blue and 100 mg hydrocortisone which was injected intradermally. After a single injection, 16 patients had relief of symptoms.

Eight of the remaining 9 patients underwent repeat injection, and 6 of these patients then achieved relief of symptoms. The overall success was 88 %. Early in the study, the authors attempted to inject under intravenous sedation only. This was abandoned in preference of general anaesthesia after 2 patients found the discomfort of injection intolerable.

The authors make an interesting technical point that may explain the mode of action of the treatment. When they observed the patients who responded to the treatment at week 2 and week 6 follow-up, the tattooing of the skin had persisted to greater than 6 weeks duration. Amongst the non-responders who had initial relief, the tattoo was present at 2 weeks but had disappeared by 6 weeks. The authors' conclusion for this was that the dye was likely to have been inadvertently injected deeply, leading to its rapid absorption and reduction of half-life and subsequent length of action on sensory nerve endings.

In their discussion, the authors address the possibility that the lidocaine or steroid may account for the symptomatic relief instead of the methylene blue. The reason given for these agents' inclusion in the injected mixture was to provide short-term symptomatic relief from pain of the injection and to reduce the perianal inflammation from the chronic irritation of pruritus. The authors come to the conclusion that the methylene blue is the active ingredient in the mixture as the lidocaine and hydrocortisone have a short half-life when compared to the longevity of the symptom relief. They do acknowledge, however, that this is unproven.

The only side effect reported in this series was a single patient who developed a transient minor faecal incontinence.

Mentes et al. report a case series of 30 patients with idiopathic pruritus ani treated by anal tattooing with methylene blue. This group did not include steroid with the methylene blue injection and also injected lignocaine alone in 6 patients prior to starting the trial. These patients all had recurrence of pruritic symptoms within 3 days and went on to successful methylene blue injection subsequently. In their series, 24 patients were symptom-free at 1 month; 5 were partial responders, 4 of whom achieved total relief with a subsequent treatment; and 1 patient did not respond at all. At 12-month follow-up, 76.7 % of patients remained symptom-free. five patients have been followed so far to 2 years and none of them have reported recurrence [14].

Sutherland et al. report the largest series of patients to undergo anal tattooing to date [16].

All patients that were referred to their unit underwent a trial of conservative management. If this failed, they were extensively worked up for secondary causes of pruritus including colonoscopy and anal mapping for underlying anorectal disorder. None of the anorectal investigations revealed pathology.

Forty-nine patients were subsequently identified with pruritus ani and underwent anal tattooing. They report that 57 % of their patients had resolution of symptoms with a total of 96 % of patients reporting significant improvement. Four of the partial responders underwent a further treatment and were rendered asymptomatic.

This group used 10 ml 1 % methylene blue mixed with 20 ml 0.5 % Marcaine with 1:200,000 epinephrine and 1 ml methylprednisolone (40 mg in 1 ml) as the initial treatment.

Sutherland reports side effects in seven patients who complained of transient faecal incontinence. This troubling symptom had resolved by 6 weeks in all patients. Two patients in their study complained that the loss of perianal skin sensation was troubling to them, and they subsequently emphasised this unavoidable (perhaps therapeutic) side effect in their consent process. This group have yet to report long-term follow-up data.

On a technical point, this group took the step of marking the symptomatic area preoperatively and ensuring that this area was completely 'inked' at the end of the procedure. Instead of linear injections along skin furrows, this group injected a series of subcutaneous 'blebs' to ensure the dye ended up in the intradermal compartment.

In summary, methylene blue tattooing seems to be a safe and dramatically successful treatment for a proportion of patients with idiopathic pruritus ani. Patients should be warned of the side effects of diminished perianal sensation that can be unpredictable in recovery: transient faecal incontinence, tattooing of the perianal skin, green-blue discoloration of the urine that lasts around 3–4 days and the possible need to repeat the procedure. Although of low risk, serious side effects

include anaphylaxis, perianal sepsis, cellulitis and skin necrosis requiring skin debridement.

It is generally recommended that the procedure be carried out under intravenous sedation or general anaesthesia as it is poorly tolerated using local anaesthesia alone.

Sutherland's technique of preoperative skin marking and subdermal bleb infiltration seems to be a sensible approach and is recommended to achieve long-term tattooing which correlates with successful outcome. A combination of steroid, local anaesthesia and methylene blue of between 0.5 and 1 % concentration should be injected. It is recommended to keep the total volume injected to less than 40 ml. Skin necrosis has been observed at a higher volume of infiltrate. Prophylactic antibiotics do not seem to be required however should be guided by local policy and operator preference.

Conclusion

Pruritus ani is uncommon but can be extremely difficult to treat. Expertise from both the dermatology and colorectal departments to identify potentially underlying disease is essential to ensure that pruritus is not a secondary symptom. When idiopathic pruritus ani has been confirmed and fails to settle with hygiene measures or stool thickeners, capsaicin treatment, if tolerated, should be instigated. If this fails or is intolerable, success may be achieved with an anal tattooing procedure using methylene blue solution which can be repeated as necessary.

References

1. Steinhoff M, Bienenstock J, Schmelz M, Maurer M, Wei E, Bíró TJ. Neurophysiological, neuroimmunological, and neuroendocrine basis of pruritus. J Invest Dermatol. 2006;126(8):1705–18.
2. Yosipovitch G, Greaves MW, Schmelz M. Itch. Lancet. 2003;361(9358):690–4.
3. Ikoma A, Steinhoff M, Ständer S, Yosipovitch G, Schmelz M. The neurobiology of itch. Nat Rev Neurosci. 2006;7(7):535–47.
4. Twycross R, Greaves MW, Handwerker H, Jones EA, Libretto SE, Szepietowski JC, Zylicz Z. Itch: scratching more than the surface. QJM. 2003;96:7–26.
5. Mazier WP. Hemorrhoids, fissures and pruritus ani. Surg Clin North Am. 1994;74:1277–92.
6. Friend WG. The cause and treatment of idiopathic pruritus ani. Dis Colon Rectum. 1977;20:40–2.
7. Daniel GL, Longo WE, Vernava III AM. Pruritus ani causes and concerns. Dis Colon Rectum. 1994;37:670–4.
8. Smith LE, Henrichs D, McCullah RD. Prospective studies on the etiology and treatment of pruritus ani. Dis Colon Rectum. 1982;25:358–63.
9. Caplan RM. The irritant role of feces in the genesis of perianal itch. Gastroenterology. 1966;50:19–23.
10. Farouk R, Duthie GS, Pryde A, Bartolo DC. Abnormal transient internal sphincter relaxation in idiopathic pruritus ani: physiological evidence from ambulatory monitoring. Br J Surg. 1994;81:603–6.
11. Dasan S, Neill SM, Donaldson DR, Scott HJ. Treatment of persistent pruritus ani in a combined colorectal and dermatological clinic. Br J Surg. 1999;86:1337–40.
12. Harrington CI, Lewis FM, McDonagh AJ, Gawkrodger DJ. Dermatological causes of pruritus ani. BMJ. 1992;305:955.
13. Bruynzeel DP. Dermatological causes of pruritus ani. BMJ. 1992;305:955.
14. Mentes BB, Akin M, Leventoglu S, et al. Intradermal methylene blue injection for the treatment of intractable idiopathic pruritus ani: results of 30 cases. Tech Coloproctol. 2004;8:11–4.
15. Botterill ID, Sagar PM. Intra-dermal methylene blue, hydrocortisone and lignocaine for chronic, intractable pruritus ani. Colorectal Dis. 2002;4:144–6.
16. Sutherland AD, Faragher IG, Frizelle FA. Intradermal injection of methylene blue for the treatment of refractory pruritus ani. Colorectal Dis. 2009;11(3):282–7.
17. Papoiu AD, Yosipovitch G. Topical capsaicin. The fire of a 'hot' medicine is reignited. Expert Opin Pharmacother. 2010;11(8):1359–71.
18. Lysy J, Sistiery-Ittah M, Israelit Y, Shmueli A, Strauss-Liviatan N, Mindrul V, Keret D, Goldin E. Topical capsaicin–a novel and effective treatment for idiopathic intractable pruritus ani: a randomised, placebo controlled, crossover study. Gut. 2003;52(9):1323–6.
19. Schirmer RH, Adler H, Pickhardt M, Mandelkow E. "Lest we forget you—methylene blue…". Neurobiol Aging. 2011;32(12):2325.e7–16.
20. Wolloch Y, Dintsman M. A simple and effective method of treatment for intractable pruritus ani. Am J Proctol Gastroenterol Colon Rectal Surg. 1979;30:34–6.
21. Eusebio EB, Graham J, Mody N. Treatment of intractable pruritus ani. Dis Colon Rectum. 1990;33:770–2.
22. Eusebio EB. New treatment of intractable pruritus ani. Dis Colon Rectum. 1991;34:289.
23. Farouk R, Lee PW. Intradermal methylene blue injection for the treatment of intractable idiopathic pruritus ani. Br J Surg. 1997;84:670.

Reconstruction of the Anus and Perianal Region

22

David Ross and Marlese Dempsey

Introduction

Acquired defects of the anus and perianal area most commonly arise due to extirpative oncologic surgery but can also follow trauma and necrotising soft tissue infection requiring extensive debridement (Table 22.1). Smaller wounds may be closed primarily or left to heal by second intention. However, larger wounds may not be primarily closed and conservative management may take several weeks or months to achieve, resulting in considerable discomfort and the distress of daily dressings. In these situations, reconstructive techniques can be employed to achieve rapid healing and restoration of anatomy and function. Numerous reconstructive options are available and must be tailored to the specific requirements of the individual patient. Small or superficial wounds may be managed with relatively simple procedures such as skin grafting or the use of local flaps. However, larger 3-dimensional defects that follow total anal resection, after abdominoperineal excision (APE) or pelvic exenteration, may produce large, complex wounds in which the aim of reconstruction may be varied.

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In addition to providing soft tissue cover, larger-volume loco-regional flaps or microvascular free flaps are required to close the wound and cover neurovascular structures and viscera. In addition, flaps will obliterate dead space and introduce well-vascularised tissue that can be particularly helpful in achieving healing in previously irradiated fields [1].

Patients are best managed in a multidisciplinary setting requiring discussion between the colorectal and plastic surgeon. Planning will also involve the expertise of other specialties, including the radiation oncologist, dietician and stoma nurse. Preoperatively patients may have had chemoradiotherapy or previous surgery, and it is therefore essential to take a careful history and examine the patient in order to avoid any potential complications due to previous scars or radiation injury.

Table 22.1 Causes of acquired anal defects

Extent of defect	Aetiology
Superficial and perianal defects	AIN/Paget's disease/melanoma and non-melanoma skin cancers
	Condyloma
	Hidradenitis suppurativa
	Pilonidal disease
	Post-debridement for trauma/infection
Partial anal defects	Local tumour excision
	Repair post-stenosis
	Inflammatory bowel disease
Total anal defects	APE
	Pelvic exenteration

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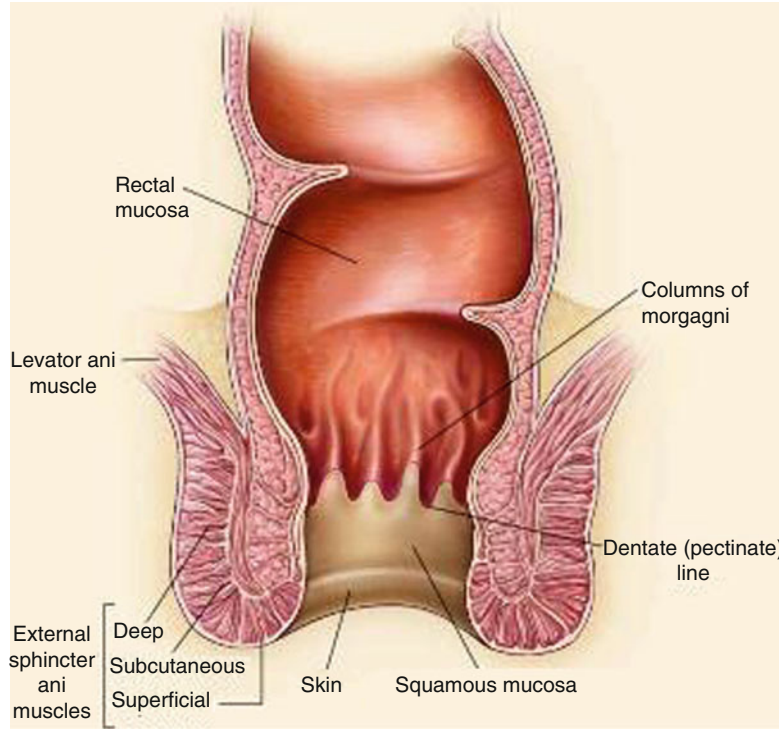

Fig. 22.1 Anatomy of the anus

Preoperative planning will include location of incisions and stomas to optimise tumour clearance and reconstructive options. Prior or intraoperative radiation and brachytherapy may also have implications for wound healing [2]. Involvement of the medical oncologist is also critical to optimise timing of surgery and minimise the deleterious effects of chemotherapy on wound healing. Several studies have shown this to occur maximally at 7–10 days after administration of chemotherapy [3, 4]. Finally, expert nursing and wound care, along with rehabilitation, will facilitate patient recovery and return to normal activities.

Anatomy

The anus can be divided into the mucosa-lined anal canal and the more distal epidermis-covered anal margin. Most of the anal canal is lined by squamous mucosa, which is present between the anal verge and the dentate (or pectinate) line. The dentate line indicates the junction of the superior part of the anal canal (derived from the embryonic hindgut) and the inferior part (derived from the embryonic anal pit or proctodeum). Proximal to this line, the anal canal is lined with columnar mucosa.

It is feasible to reconstruct the perianal area and anal canal to the level of the dentate line with cutaneous tissue (Fig. 22.1), and several options will be described in more detail below. This chapter will also review the techniques available to repair defects following proctectomy.

The perineum has a rich blood supply with multiple perforating vessels. Anatomically, there are encircling anastomoses around any orifice or joint. The perineum has two outlets: the urogenital and the anal. The arterial network of this region is supplied by vessels of the lower abdomen, medial thigh and gluteal region [5]. Knowledge of the rich blood supply can be applied to the design and elevation of several flaps valuable in perianal reconstruction.

Reconstructive Principles

In the first instance it is essential to gain a clear understanding of the extent of the defect and its composition. This will allow decisions to be made with regard to what is likely to be required to restore both structure and function. For an anal defect, the wound needs to be assessed in terms of loss of cover, support and lining. Reconstruction of each component requires careful consideration in order to select the appropriate tissue replacement and, ideally, to replace 'like with like'. In this way an optimal functional (and aesthetic) result can be achieved. As a general rule, perianal defects can be repaired using simple local or fasciocutaneous flaps to provide thin pliable skin to resurface the area required and contour. Flaps will also need to be robust and able to contour into the dentate line.

Surgical planning will also help to achieve tension-free closure, control dead space and minimise patient and donor-site morbidity. The latter is of course important, as a poorly healing perineal wound will prove to be very upsetting to any patient, as well as limit mobilisation and independence. As noted above, the ability to achieve uncomplicated wound healing will be particularly challenging in previously irradiated areas. However, care in planning and execution can reduce the risk of fistulae and sinus formation and minimise downtime. In addition, thought should also be given to potential 'backup' methods in the event of failure and the need for salvage reconstruction.

Preoperative Planning

Classification of Defects

When planning reconstruction it can be helpful to classify the extent of an existing or planned defect and its associated reconstructive options. Below, we have proposed a simple classification system that divides defects into superficial partial and total anal defects. Superficial partial defects are confined to the perianal skin and subcutaneous tissues and commonly follow excision of AIN, Paget's disease and other cutaneous malignancies. Surgical removal of these lesions may result in defects that involve resection of deeper tissues, including the internal anal sphincter.

We have classified superficial perianal defects on the extent of the defect; a type 1 resection will involve less than one third of the anal margin, a type 2 defect will comprise between one and two thirds of the circumference, and a type 3 defect is seen following loss of between two thirds and the total circumference of the anal margin. We find that this classification system can simplify the preoperative decision-making process and can be used as an algorithmic approach to reconstructive planning (Fig. 22.2).

Role of Faecal Diversion

As a general rule, it was accepted that patients undergoing some form of anal margin resection may be best served with a covering stoma in order to reduce both trauma on the repair and the risk of infection. Creation of a stoma is always going to be stressful and upsetting for a patient, even if only temporary. However, for all but type 3 defects, it has also proven to be unnecessary and associated with considerable morbidity. The additional effects of a stoma include skin irritation (12 %), pain associated with poor stoma location (7 %) and partial necrosis (5 %) [6].

Our experience has, therefore, led us to try and avoid a de-functioning stoma whenever possible (Fig. 22.3). In patients with smaller resections, we will prepare the bowel on the day prior to surgery using two sachets of Citramag. Thereafter, patients are placed on a liquid/low-residue diet and encouraged to open their bowels as per normal. A normal diet is then reintroduced at 5 days thereafter.

Principles of Anal Reconstruction

Reconstructive methods follow a 'reconstructive ladder' (Fig. 22.3), beginning with primary closure and progressing to free tissue transfer at the other end of complexity. Ascent of this ladder

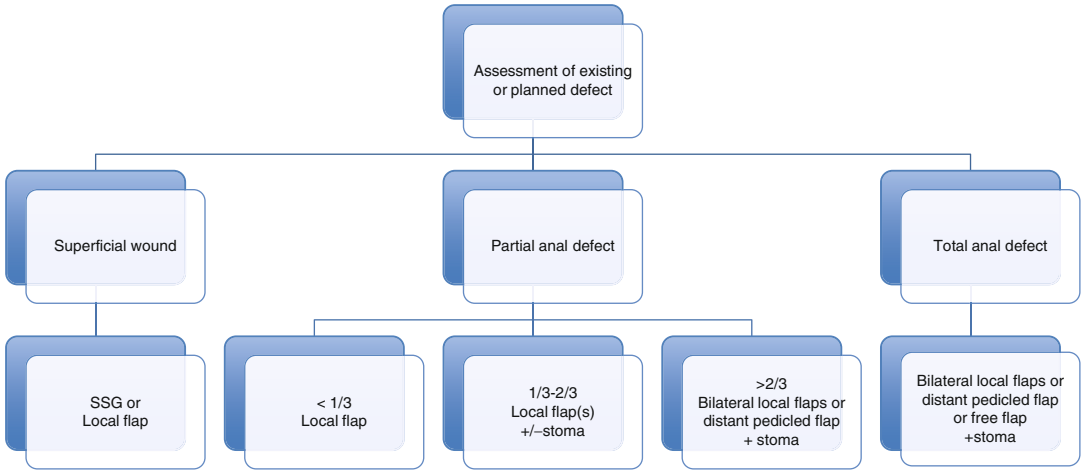

Fig. 22.2 Anal reconstructive algorithm

does not always imply a superior result as outcome depends on the appropriate selection of the right technique for the wound or defect.

Skin Grafts

A skin graft consists of epidermis and some or all of the dermis. By definition, a graft is something that is removed from the body, de-vascularised and transferred to another location [7]. Grafts of any kind require vascularisation from the recipient bed onto which they are placed for survival. Therefore, grafts require a stable, well-vascularised bed in order to 'take'.

Split-thickness skin grafts (SSG) contain epidermis and varying amounts of dermis. The donor area heals by re-epithelialisation that derives from the residual dermis. These grafts are relatively simple and safe to harvest and provide a plentiful source of soft tissue cover. However, they are thin, vulnerable to trauma and prone to contraction.

Full-thickness skin grafts (FSG) contain epidermis and the entire dermis, so only limited quantities are available if the donor sites are to be closed primarily. They are usually harvested from the groin or abdominal area. Compared to SSG they provide a better texture match and are less susceptible to contraction. However, they are less plentiful and heal more slowly than SSG. They are also more likely to fail due to their thickness.

Skin grafts have a limited role in resurfacing the perianal area as they are challenging to stabilise and vulnerable to infection and movement. In addition they tend not to 'take' upon a previously irradiated wound bed and will not survive on bare bone or a sloughy, infected wound. Furthermore, once split skin grafts have become established, contraction can retract and distort the local anatomy to produce functional, aesthetic and psychological sequelae [8].

Flaps

Flap-based techniques provide the mainstay of perianal reconstruction and are used to repair the majority of defects in this area. Flaps are intrinsically different to grafts due to the fact that these tissue composites retain their blood supply. *A flap is defined as a unit of tissue of variable composition which, when transferred from a donor to a recipient site, brings its own blood supply and intrinsic circulation* [9]. Flaps can be further classified in a number of ways, but the three most significant characteristics are blood supply, movement and composition.

Flaps are perfused by either a random or axial vascular pattern (Fig. 22.4). Random pattern flaps have no discernible dominant blood supply

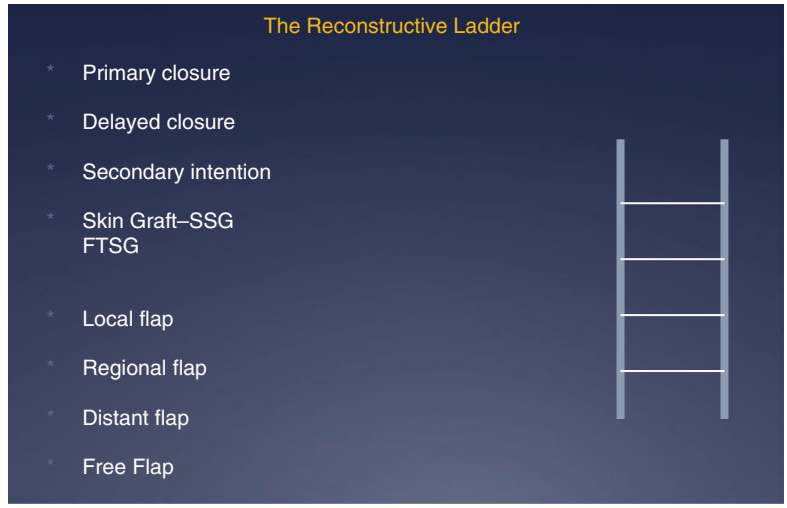

Fig. 22.3 The reconstructive ladder

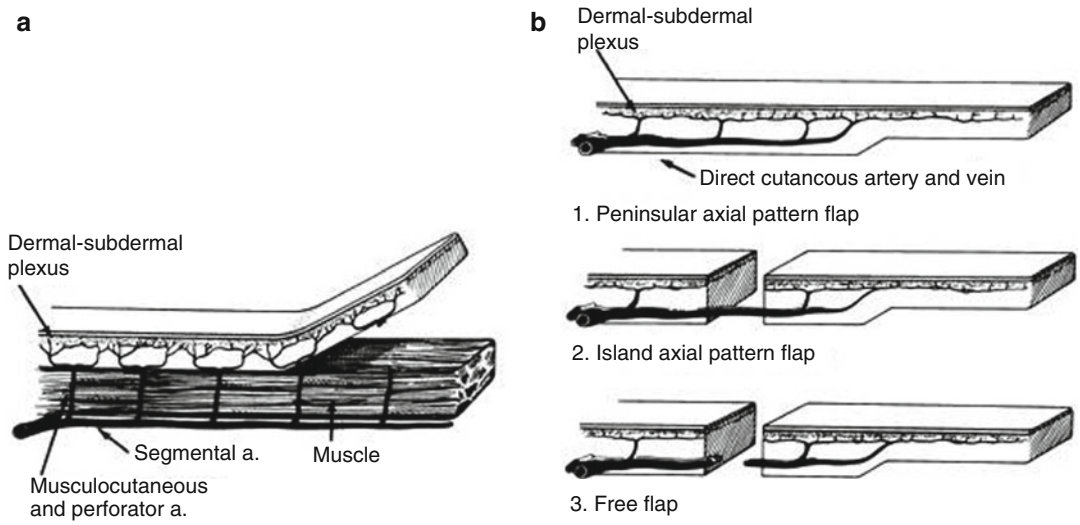

Fig. 22.4 Flap type and blood supply. (**a**) Random pattern skin flap. (**b**) Axial pattern skin flaps

and are usually fed by a dermal-subdermal plexus. Axial pattern flaps are perfused by an identifiable, single arteriovenous system that proceeds along the longitudinal axis of the flap. As a consequence, a larger and more reliable flap can be raised when there are identifiable blood vessels within the tissues.

Flaps can also be classified by their positional relationship to the defect, i.e. as local, distant or free flaps. Local flaps utilise tissues adjacent to the defect that may be advanced along the axis of the flap (e.g. V-Y). Alternatively they may be rotated, transposed or interpolated (i.e. passed over or under an intervening tissue bridge to reach the defect). Local flaps are perfused by a random pattern blood supply and tend to be used for smaller defects. As a result they bring in skin of similar quality, i.e. thickness, elasticity and, where appropriate, colour.

The perianal region is supported by a rich supply of loco-regional plexuses that allow design and harvest of multiple different local flaps. Some of these can be thin and pliable, consisting of skin and fascia alone, such as the ‘lotus petal’

and 'Singapore' flaps. More substantial local perforator flaps, including the inferior gluteal perforator flap (I-GAP), provide greater volume and area of skin cover and soft tissue to fill dead space. Distant flaps, as their name implies, are raised from sites away from the resection and may be transferred either by use of a pedicle (i.e. the rectus abdominis flap) or as free tissue transfers by disconnection and re-vascularisation using microsurgical techniques. Flap composition is not limited to skin or subcutaneous tissues alone but may comprise any tissue or combination of tissues, to include skin, fat, muscle, fascia, bone and tendon.

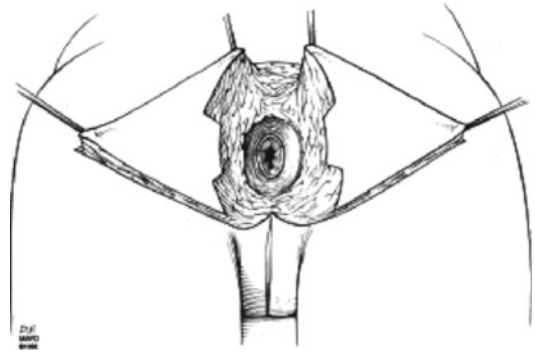

Fig. 22.5 V-Y flap

Routine Flaps for Anal and Perianal Reconstruction

As noted above, flaps usually provide the best method of reconstructing anal and perianal defects as they are composed of robust, well-vascularised tissue. The flaps listed below represent a partial, though not exhaustive, list of those that should be considered preoperatively.

Local Flaps

The extensive vascular supply of the perineal, gluteal, infragluteal and upper thigh regions allows for many regional fasciocutaneous flaps to be raised. Most of these flaps may be applied as either V-Y flaps, pedicled transposition flaps, subcutaneously pedicled flaps or true island flaps. Because the vascular systems of the perineal and gluteal regions anastomose with each other, these flaps may be raised as extended flaps or as reversed flaps depending on retrograde blood flow.

Random Pattern Flaps

The *V-Y flap* (Fig. 22.5) is a type of local advancement flap that moves forward from donor to recipient site without rotation or lateral movement. This flap is based upon subcutaneous vessels that usually perforate from small intramuscular vessels emanating from below. The laxity in the subcutaneous tissues permits advancement and lateral elasticity in the skin permits closure of the tail (secondary defect). These flaps have proven to be particularly effective in reconstructing defects following superficial anal malignancies and perianal cutaneous cancers [10].

V-Y flaps are also of great value in repairing unhealed perineal and perianal wounds (Fig. 22.6) and can be adapted as a 'diamond'-shaped flap to address post-surgical anal stenosis (Fig. 22.7).

The *rhomboid flap* was first described by Limberg and uses a simple geometric design to close defects with local tissue (Fig. 22.8). It can be used to close type 1, partial anal margin defects (see Fig below), and is routinely employed to reconstruct natal cleft defects following wide excision for pilonidal sinus [11]. Figure 22.9 shows use of a rhomboid flap to repair a type 1 perianal defect following excision of localised Paget's disease.

Local flaps can be combined to repair larger type 2 and 3 defects. Figure 22.10 shows an extensive defect following excision of recurrent AIN3. The defect was closed with a combination of a sliding transposition flap and a V-Y flap on the opposite side. By rotating the inferior flap, linear scarring above and below the anal margin was avoided, thus limiting the risk of post-surgical stenosis.

Perforator Flaps

Fasciocutaneous flaps based on the terminal branches of the superficial perineal artery remain 'workhorse' flaps in perianal reconstruction. Multiple flaps have been described effectively

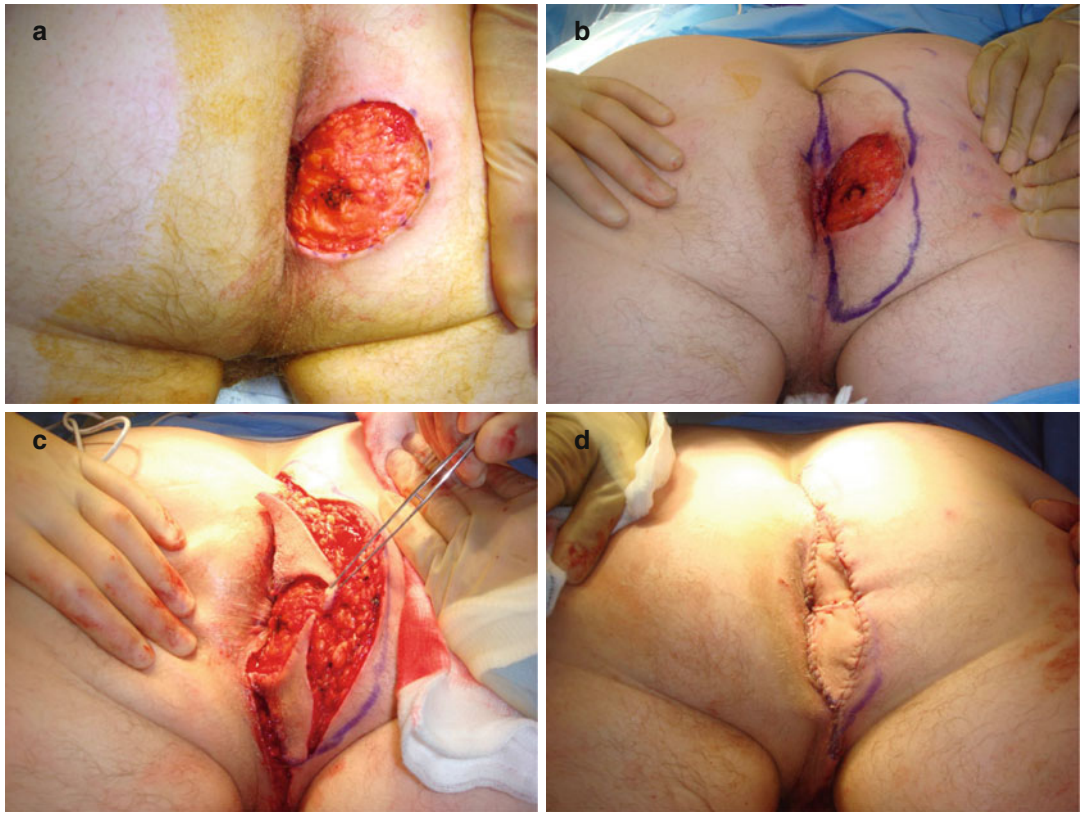

Fig. 22.6 (**a**) Type 1 defect, involving approximately one-third of the right peri-anal margin following resection of AIN3. (**b**) Planned reconstruction using bilateral opposing V-Y flaps. (**c**) Flaps elevated prior to insetting along the anal margin. (**d**) Flaps inset and anal margin resurfaced

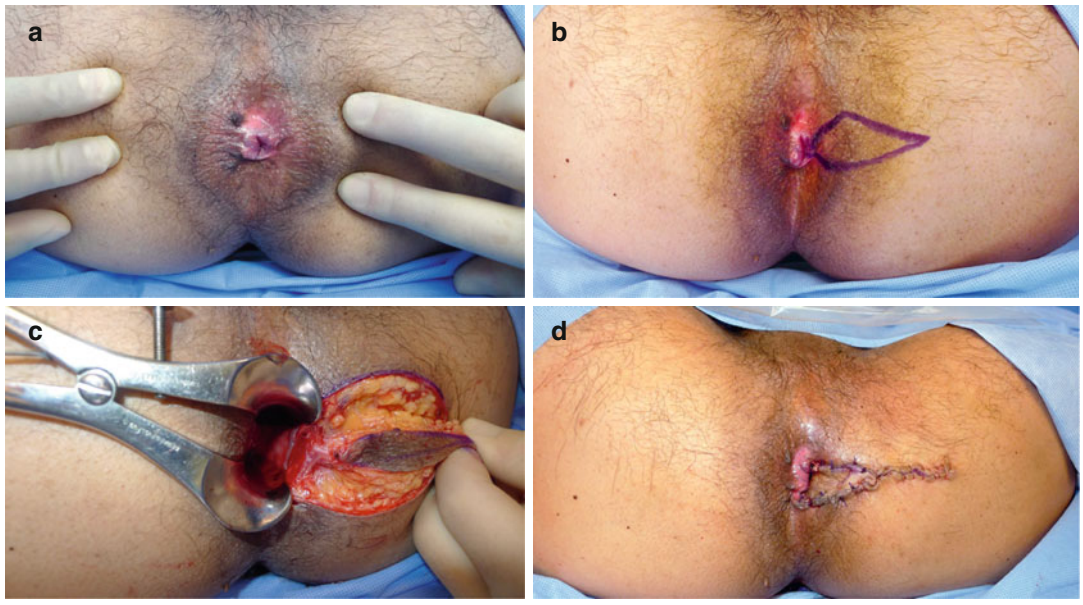

Fig. 22.7 (**a**) Anal stenosis post-hemorroidectomy. (**b**) Flap outline. (**c**) Flap elevation. (**d**) Flap inset

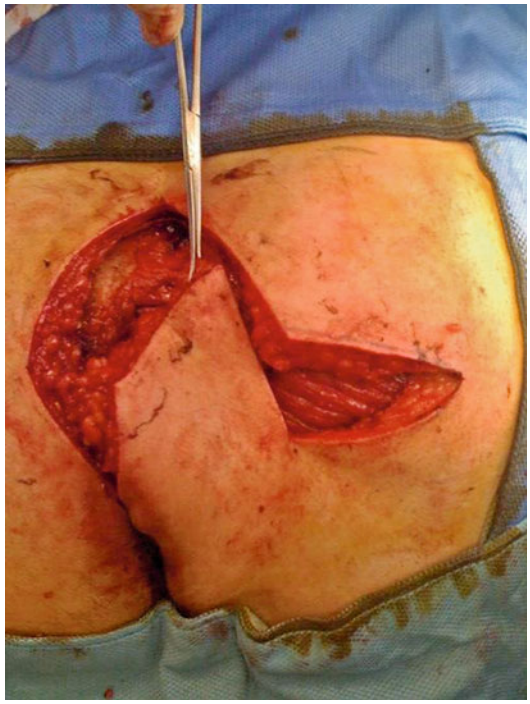

Fig. 22.8 Rhomboid flap; the flap has been transposed to close the defects

utilising the same source of blood supply but with different anatomic supports. These flaps are sited over a dense area of perforators with a rich anastomotic network that lies just anterolateral to the anus. The 'lotus petal' flap is a fasciocutaneous flap that can be harvested from the gluteal fold. Once elevated the flap can be rotated around an arc similar to the leaves of the lotus flap, providing a flexible and adaptable source of tissue for loco-regional reconstruction [12].

The *neurovascular pudendal thigh flap* was described by Wee and Joseph from Singapore in 1989 [13] and modified by Woods et al. in 1992 [14]. This fasciocutaneous flap is based on the posterior labial vascular bundle and innervated by the posterior labial branch of the pudendal nerve. It can provide a long and well-vascularised sensate flap that is thin and pliable (Fig. 22.10). Both the 'lotus' and 'Singapore' flaps are of considerable use in reconstructing perianal and vulval defects as they provide thin, pliable tissue that is robust. However, they are also noteworthy for producing minimal donor-site morbidity.

Distant Flaps

Rectus Abdominis Myocutaneous Flap

Myocutaneous flaps from the abdomen are the most common source of tissue used to reconstruct significant perineal defects and to fill dead space. The rectus abdominis myocutaneous (RAM) flap, described by Taylor et al., consists of a large skin paddle based upon the underlying rectus abdominis muscle and vascularised by the deep inferior epigastric vessels. This allows the flap to be passed behind the pelvis to reach the perineum [15]. This flap has several advantages; it may be moved around a wide arc of rotation based on the consistent deep inferior epigastric artery pedicle. This tissue source can provide considerable tissue bulk, particularly when combined with an overlying skin paddle, and is reasonably robust [16]. The RAM flap can be designed in a variety of modifications, depending on the extent of the defect. In most cases it is raised as a vertical rectus abdominis flap (VRAM). By modifying the skin island to an oblique paddle, a larger area of skin can be harvested in order to reconstruct larger defects.

The flap is raised on the contralateral side to the planned stoma; the laparotomy incision should place the umbilicus on the side of the preserved rectus to prevent necrosis.

In many centres the VRAM remains the method of choice to repair the post-APE defect. However, there are important limitations to this choice that have demoted its use in our unit, behind that of the I-GAP flap. Specifically, the VRAM requires harvest of a muscle necessary to the integrity and function of the abdominal wall and is an important adjunct to the creation of a stoma. The donor site can be problematic if a large skin island is required in either the thin or overweight patient, leading to potential hernia formation. It is also counterintuitive to use a VRAM if the bowel has been endoscopically mobilised as part of a laparoscopic APE. Once the flap has been mobilised and islanded on its pedicle, it tends to atrophy, and thus, bulk to obliterate dead space is lost. Traction on the pedicle can result in partial or flap loss, and finally, the sacrifice of a potential future new stoma or urostomy site.

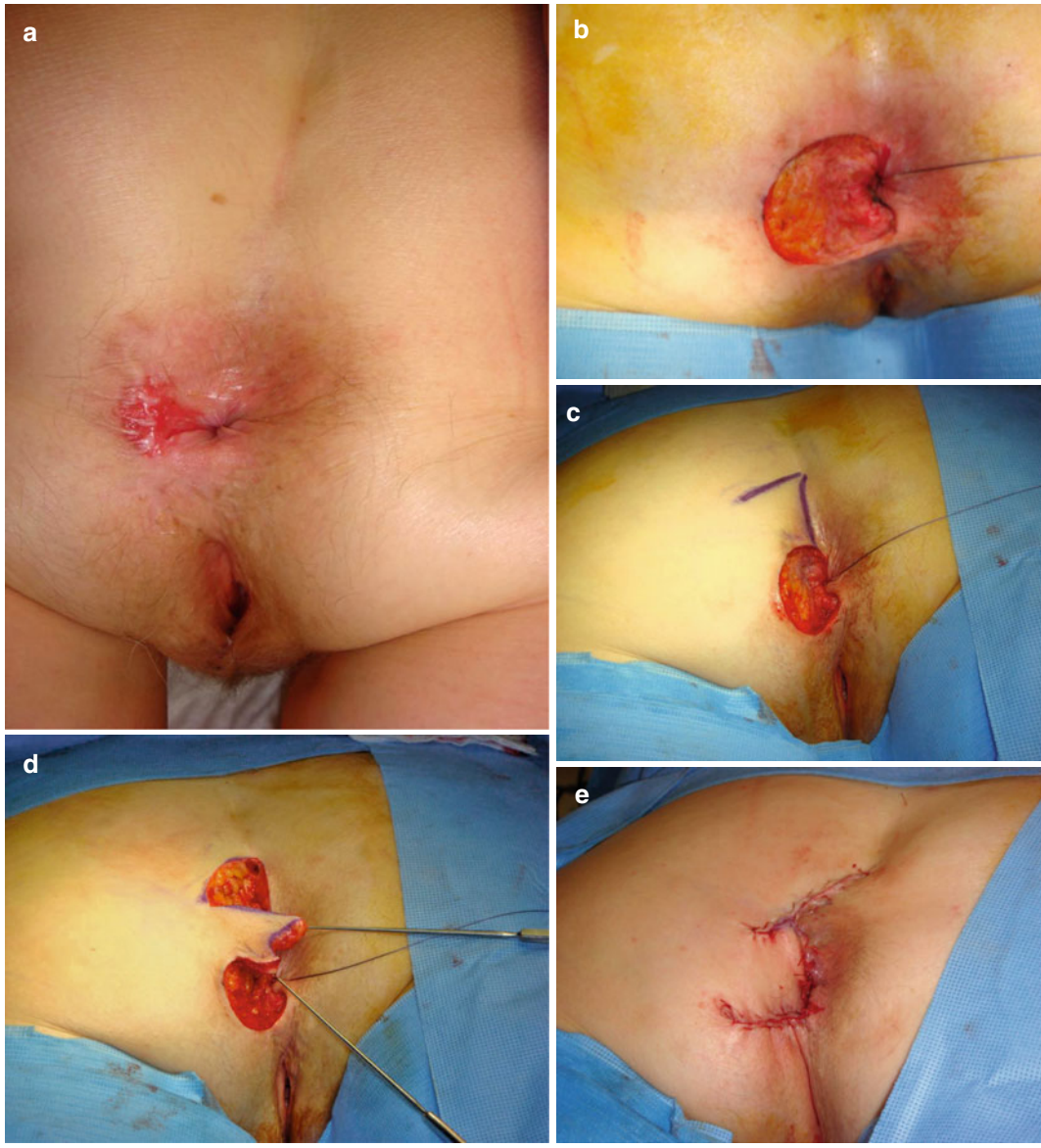

Fig. 22.9 (**a**–**e**) Repair of type 2 defect post resection of AIN 3 using a rhomboid flap. (**a**) Pre-excision view showing the right peri-anal lesion. (**b**) The excision defect. (**c**) Flap planning. (**d**) Flap elevated. (**e**) Flap inset – no de-functioning stoma was required

Gluteal Perforator Flaps

Holm advocated the use of bilateral *gluteal musculocutaneous flaps* to aid primary closure following APE [17]. However, this involves significant muscle injury with its attendant problems of pain and delayed mobility. Since 2007, the *inferior gluteal artery perforator* (*I-GAP*) *flap* has become our technique of choice in providing soft tissue to repair hemi and total anal resection defects, particularly following APE in the immediate and delayed setting. This flap utilises the same skin and subcutaneous tissue as the gluteus myocutaneous flap but avoids the need to use or injure any underlying muscle. It was described as a pedicled flap for coverage in ischial and sacral pressure sores and can be islanded on its perforators to rotate and cover multiple local defects. It has become our method

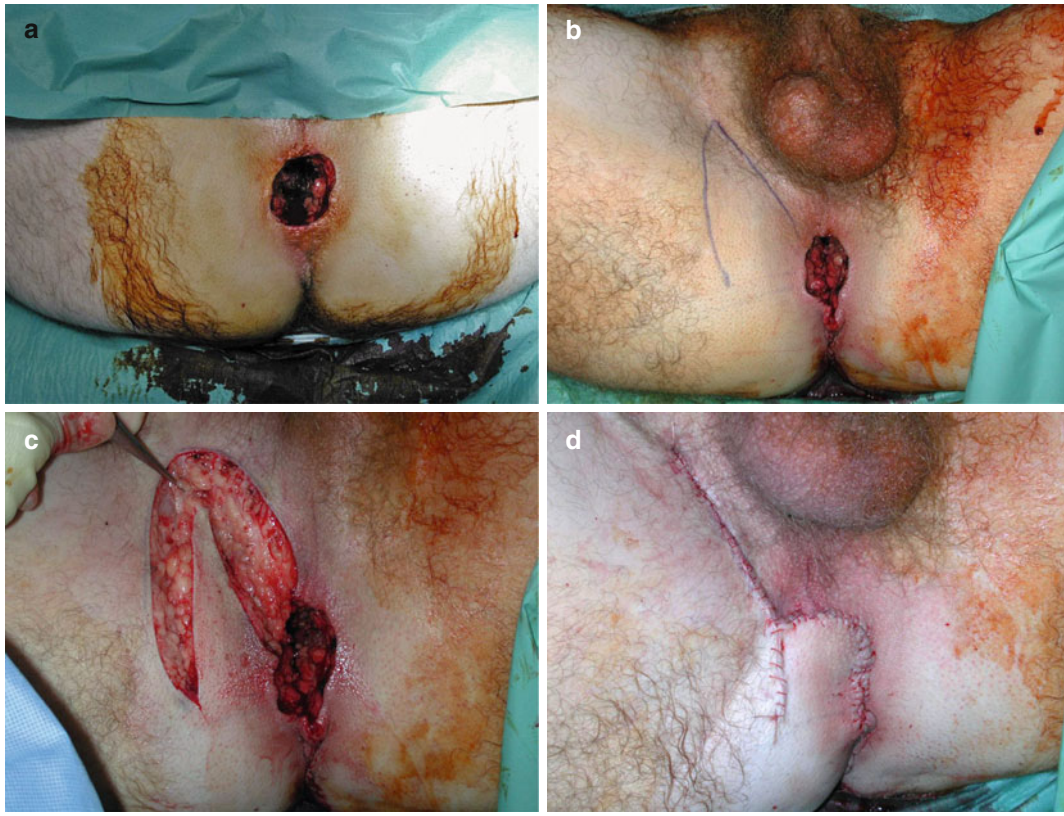

Fig. 22.10 (**a**) Defect following excision of peristent Crohn's sinus. (**b**) Planning of the "Singapore" flap. (**c**) Flap elevation. (**d**) The flap has been de-epithelialised in its distal half and the tissue buried within the defect to provide healthy vascularised tissue in order to achieve rapid healing. Note the excellent and hidden donor site closure

of choice when repairing type 3 perianal defects and total anal resurfacing. Bilateral flaps are advanced into the defect and down onto the dentate line (Fig. 22.11). These flaps should be long and as wide as required to ensure adequate tissue harvest but also the possibility of further flap advancement if further resection is required. We have found the use of a 'Lone Star' retractor to be very valuable in visualising the defect and facilitating the repair. It is advisable to introduce radial 2/0 Vicryl sutures into the residual anal wall in order to 'parachute' and secure the flaps into an optimum location. The main complication after surgery to resurface most, or all of the anal margin, has been post-surgical stenosis (20 %), requiring self-dilatation and recurrent disease (30 %). Patients undergoing type 3 resection for extensive AIN require careful post-surgical surveillance, and as noted above, a significant proportion have gone on to require completion APE.

The I-GAP has also proven itself to be exceptionally useful in repairing the post-APE defect (Fig. 22.12). The stable tissue bulk (comprising skin, fat and fascia only) allows effective and lasting control of dead space and the introduction of well-vascularised tissue to facilitate closure of the large, previously irradiated wounds that accompany APE. We have used this flap to reconstruct perineal defects with encouraging success [18]. This flap has low donor-site morbidity in comparison to its alternatives, allows early mobilisation and can be shaped to provide vaginal reconstruction where necessary. As a consequence, we have been able to mobilise patients undergoing APE earlier and reduce the duration of hospital admission. This technique has proven to be exceptionally valuable in treating patients with an unhealed perineum after cancer excision. It can also be effective in dealing with the post-proctectomy patient with inflammatory bowel disease, though

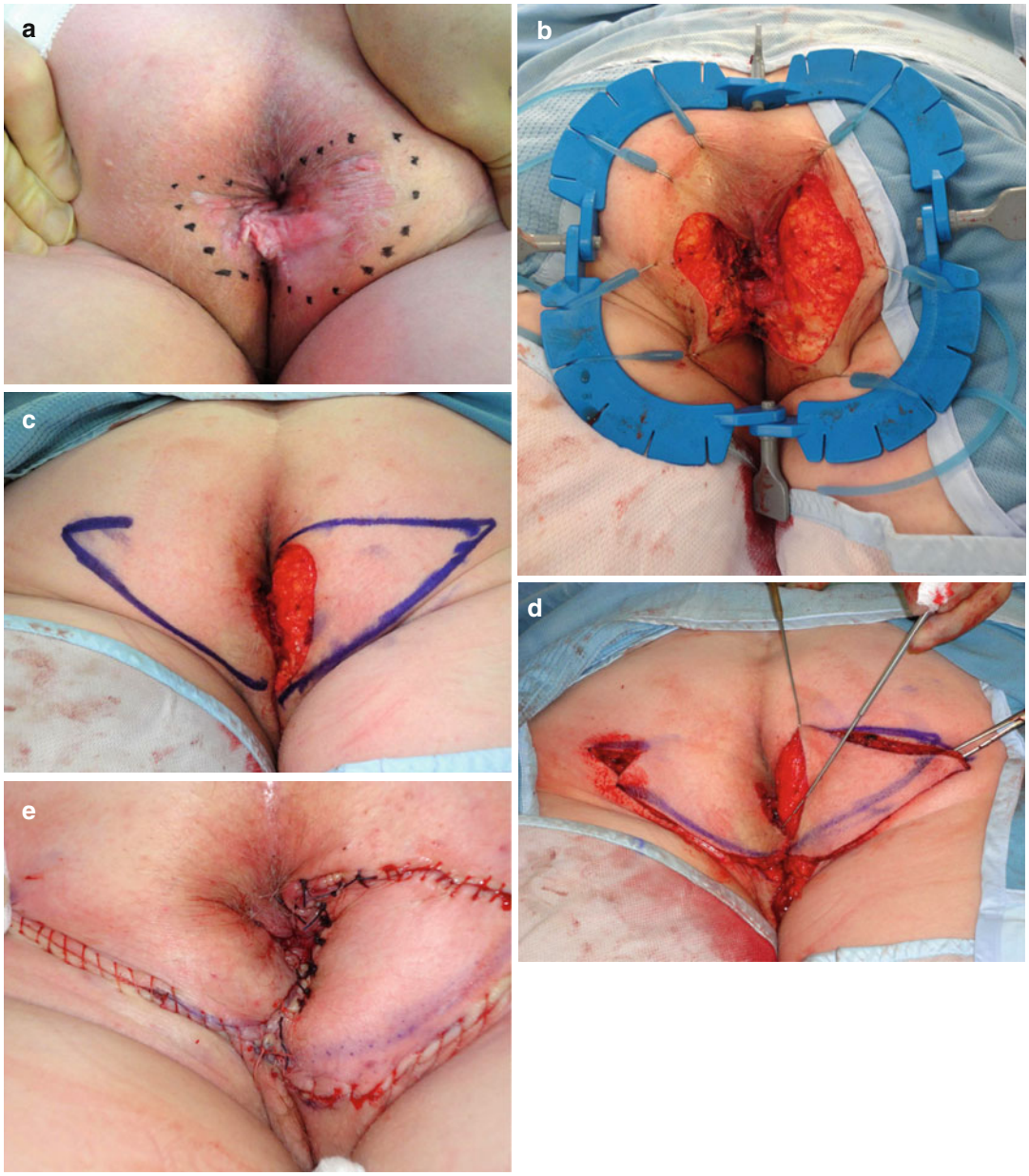

Fig. 22.11 (**a**) Type 3 perianal defect; *Dotted line* shows proposed excision margin for extensive AIN3. (**b**) Excision defect (type 3). (**c**) Proposed flap reconstruction; The right side has been planned as a V-Y advancement flap. The left side flap is a sliding transposition flap. (**d**) Flaps elevated. (**e**) Flaps inset and wounds closed with an absorbable (vicryl) suture

healing can remain problematic in these patients due to the nature of their underlying pathology.

Posterior Thigh Fasciocutaneous Flap

The posterior thigh fasciocutaneous flap is a reliable flap for providing closure of the perianal region. It represents a reliable alternative to muscle-based flaps, as minimal functional deficit will result. The flap is based on the descending branch of the inferior gluteal artery, which provides an excellent source of sensate coverage [19–21].

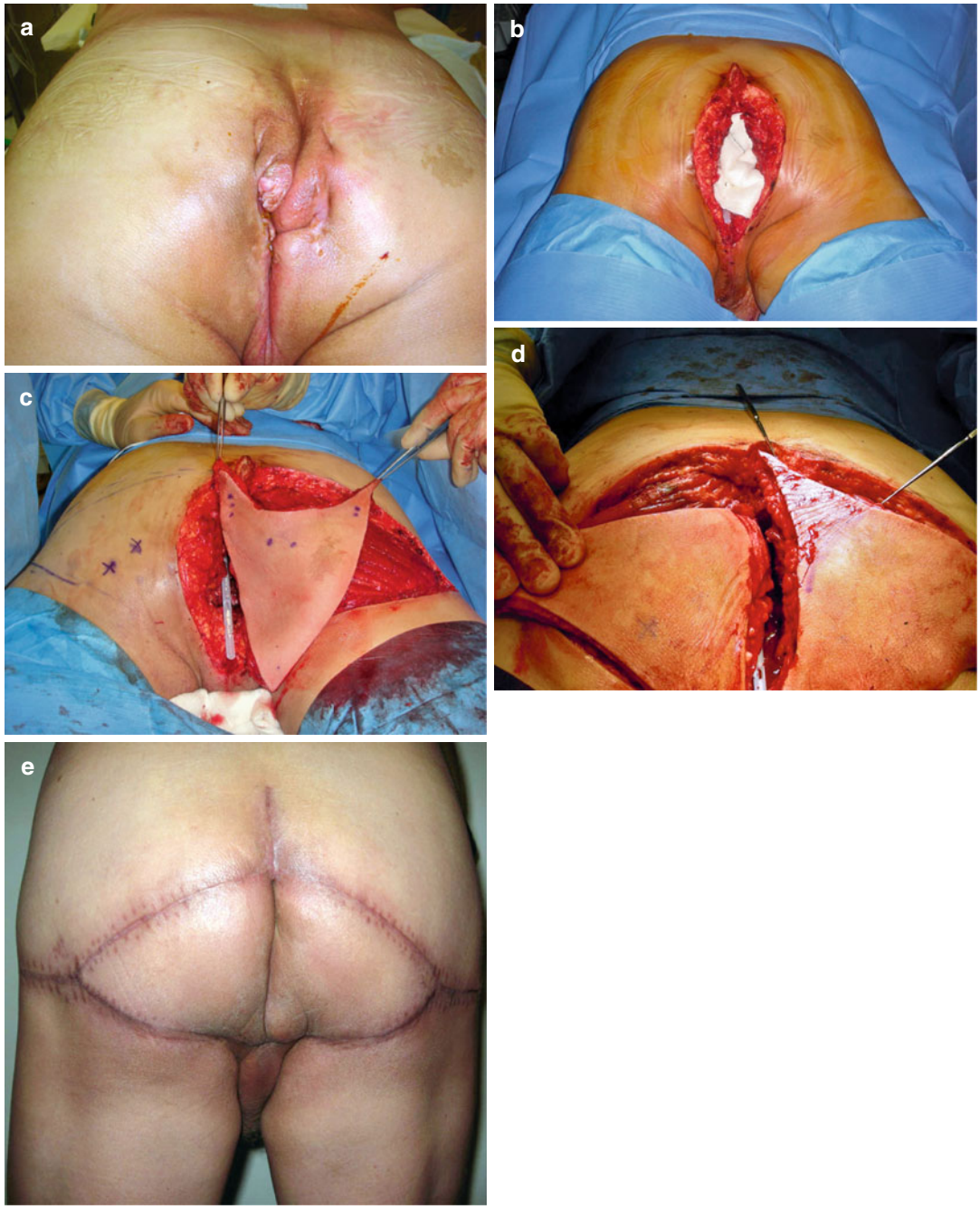

Fig. 22.12 (**a**) This shows a recurrent Buschke-Lowenstein tumour following previous radiotherapy that required abdominoperineal excision (APE). The following views show reconstruction to the defect using bilateral inferior gluteal artery perforator flaps. (**b**) The APE defect. (**c**) Bilateral flaps are planned to close the defect. The right I-GAP flap has been elevated on its pedicle. Note that the *blue marks* that identify the site of vascular peforators identified with a hand-held doppler prior to elevation. The right I-GAP has been elevated and is shown rotated on its pedicle. (**d**) Both flaps are elevated and the medial section of each flap medialised prior to advancing the flaps into the deep and wide defect. In this way, the wound is closed and dead space controlled. (**e**) Stable and well healed flaps at 3 months post APE. Of note this patient returned to cycling to work at this time

Anterolateral Thigh Flap

This flap was first reported by Song et al. in 1984 [22]. Wang et al. described the use of the anterolateral thigh flap for perineal reconstructions, and it has subsequently been used widely to reconstruct defects both as a free and pedicled flap [23]. The anterolateral thigh flap is versatile and can provide a large, thin skin flap based on a single suitable fasciocutaneous or musculocutaneous perforator. It is gaining in popularity due to its excellent donor site that can, depending on the size of the flap, be primarily closed.

Functioning Muscle Flaps

Efforts to reinforce the sphincter muscles in children using the gluteus maximus muscle flap were first described over 100 years ago [24]. The procedure was revised in the first half of the last century in an attempt to improve the outcome. However, it was eventually superseded by the graciloplasty. The use of the gracilis muscle flap for anal sphincter reconstruction was first described in 1952 [25]. As opposed to the bulky muscle belly of the gluteus requiring bilateral transposition to encircle the anus, the gracilis is a long, flat muscle belly that allows unilateral mobilisation to complete the wrap. In 1988 this technique was further advanced by the connection of a pulse generator to a patient with suboptimal function of the gracilis muscle transposition. The result was a neo-sphincter with involuntary resting tone [26]. This form of dynamic graciloplasty can involve two or three phases and may be dependent on the temporary use of a stoma; however, it has proved to be an oncologically sound procedure with an appreciable degree of continence in most patients [27].

Microvascular Flaps

Microsurgery has increased the repertoire of flaps for reconstruction. As the number of options in loco-regional and pedicled flaps in perineal reconstruction is large, the indications for microsurgical reconstructions are limited to repair very large defects. These would tend to follow massive post-traumatic repair and following hemipelvectomy.

Conclusion

Surgical excision of part or all of the anal region can result in difficult wounds that may be slow to heal, or indeed never do so, particularly following neo-adjuvant chemoradiotherapy. As with optimal management of all complex or malignant disease, teamwork that brings together multiple disciplines can only help the patient achieve a better overall result.

Plastic surgeons have a significant contribution to make to the surgical care of both primary and post-surgical perineal and perianal wounds and ideally should be involved at an early stage in the decision and treatment pathway. There are an extensive range of local and regional flaps that can be utilised to achieve rapid healing of wounds and protection of underlying vital structures. Skin resurfacing can be achieved without recourse to a stoma for partial resection, though will be required for 3–6 months following larger anal resections and repair. Future challenges remain in ensuring colorectal teams gain adequate plastic surgical support, but the evidence suggests this trend is changing as the benefits become more evident and techniques evolve.

References

1. Mathes SJ, Hurwitz DJ. Repair of chronic radiation wounds of the pelvis. World J Surg. 1986;10(2):269–80.
2. del Carmen MG, Eisner B, Willet CG, Fuller AF. Intraoperative radiation therapy in the management of gynecologic and genitourinary malignancies. Surg Oncol Clin N Am. 2003;12(4):1031–42.
3. Stiernberg CM, Williams RM, Hokanson JA. Influence of cisplatin on wound healing – an experimental model. Otolaryngol Head Neck Surg. 1986;95(2):210–2.
4. Mullen BM, Mattox DE, Von Hoff DD, Hearne EM. The effect of preoperative adriamycin and dihydroxyanthracenedione on wound healing. Laryngoscope. 1981;91(9 Pt 1):1436–43.
5. Niranjan N. Perforator flaps for perineal reconstructions. Semin Plast Surg. 2006;20:133–44.

6. Park JJ, Del Pino A, Orsay CP, Nelson RL, Pearl RK, Cintron JR, Abcarian H. Stoma complications: the Cook County Hospital experience. Dis Colon Rectum. 1999;42(12):1575–80.
7. Thorne C, Grabb WC, Smith JW. Grabb & Smith's plastic surgery. 6th ed. Philadelphia: Lippincott Williams & Wilkins; 2007.
8. Hage JJ, Beurden M. Reconstruction of acquired perineovulvar defects: a proposal of sequence. Semin Plast Surg. 2011;25(2):148–54.
9. Giele H, Cassell O. Plastic and reconstructive surgery. Oxford: Oxford University Press; 2008.
10. Wietfeldt ED, Thiele J. Malignancies of the anal margin and perianal skin. Clin Colon Rectal Surg. 2009; 22(2):127–35.
11. El-Tawil S, Carapeti E. Use of a double rhomboid transposition flap in the treatment of extensive complex pilonidal sinus disease. Colorectal Dis. 2009; 11(3):313–7.
12. Yii NW, Niranjan NS. Lotus petal flaps in vulvovaginal reconstruction. Br J Plast Surg. 1996;49(8): 547–54.
13. Wee JT, Joseph VT. A new technique of vaginal reconstruction using neurovascular pudendal-thigh flaps: a preliminary report. Plast Reconstr Surg. 1989;83(4):701–9.
14. Woods JE, Alter G, Meland B, Podratz K. Experience with vaginal reconstruction utilizing the modified Singapore flap. Plast Reconstr Surg. 1992;90(2):270–4.
15. Taylor GI, Corlett RJ, Boyd JB. The versatile deep inferior epigastric (inferior rectus abdominis) flap. Br J Plast Surg. 1984;37(3):330–50.
16. Butler CE, Gundeslioglu AO, Rodriguez-Bigas MA. Outcomes of immediate vertical rectus abdominis myocutaneous flap reconstruction for irradiated abdominoperineal resection defects. J Am Coll Surg. 2008;206(4):694–703.
17. Holm T, Ljung A, Haggmark T, Jurell G, Lagergren J. Extended abdominoperineal resection with gluteus maximus flap reconstruction of the pelvic floor for rectal cancer. Br J Surg. 2007;94(2):232–8.
18. Hainsworth A, Al Akash M, Roblin P, Mohanna P, Ross D, George ML. Perineal reconstruction after abdominoperineal excision using inferior gluteal artery perforator flaps. Br J Surg. 2012;99(4): 584–8.
19. Hurst RD, Gottlieb LJ, Crucitti P, Melis M, Rubin M, Michelassi F. Primary closure of complicated perineal wounds with myocutaneous and fasciocutaneous flaps after proctectomy for Crohn's disease. Surgery. 2001;130(4):767–72; discussion 772–63.
20. Fujii M, Terashi H, Hashikawa K, Tahara S. Functional reconstruction of large anogenital defects with bilateral V-Y advancement posterior thigh fasciocutaneous flaps. J Plast Reconstr Aesthet Surg. 2007;60(11): 1208–13.
21. Kishi K, Nakajima H, Imanishi N, Nakajima T. Anal and perianal reconstruction after extramammary Paget's disease using a posterior thigh flap with a thin square wing. J Plast Reconstr Aesthet Surg. 2010;63(8):1353–6.
22. Song YG, Chen GZ, Song YL. The free thigh flap: a new free flap concept based on the septocutaneous artery. Br J Plast Surg. 1984;37(2):149–59.
23. Friji MT, Suri MP, Shankhdhar VK, Ahmad QG, Yadav PS. Pedicled anterolateral thigh flap: a versatile flap for difficult regional soft tissue reconstruction. Ann Plast Surg. 2010;64(4):458–61.
24. Chetwood CH. Plastic operation of the sphincter ani with report of a case. Med Rec. 1902;61:529.
25. Pickrell KL, Broadbent TR, Masters FW, Metzger JT. Construction of a rectal sphincter and restoration of anal continence by transplanting the gracilis muscle; a report of four cases in children. Ann Surg. 1952; 135(6):853–62.
26. Cera SM, Wexner SD. Muscle transposition: does it still have a role? Clin Colon Rectal Surg. 2005; 18(1):46–54.
27. Dev VR, Gupta A. Plastic and reconstructive surgery approaches in the management of anal cancer. Surg Oncol Clin N Am. 2004;13(2): 339–53.

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