Endoanal Ultrasonographic Imaging of the Anorectal Cysts and Masses

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Learning Objectvies

1. To understand the role of 3D-ultrasonography in the assessment of endometriosis
2. To understand the role of 3D-ultrasonography in the assessment of anorectal cysts
3. To understand the role of 3D-ultrasonography in the assessment of anorectal masses

10.1 Introduction

Anorectal cysts, endometriosis of the rectovaginal septum, and non-mucosal rectal lesions (rare neoplasias of the muscularis propria, connective tissues, and vascular structures) require imaging for proper case management. At present, endovaginal ultrasonography and magnetic resonance have become important parts of diagnostic workup of these lesions. This chapter is devoted to discussing the emerging role of three-dimensional endoanal (3D-EAUS) and endorectal (3D-ERUS) ultrasonography particularly with regard to the advantages of these techniques in evaluating the invasion of the rectal layers and adjacent organs.

10.2 Endometriosis

Endometriosis is defined by the presence of endometrial glands and stroma outside the endometrial cavity and the myometrium. The most common locations of the ectopic endometriotic implants are found in the pelvis (ovaries and pelvic peritoneum) and followed by deep infiltration sites (uterosacral ligaments, rectosigmoid colon, vagina, and bladder). Imaging techniques have been recommended for the diagnosis and identification of the lesion location [1, 2]. Several reports have demonstrated the accuracy of ultrasonography, performed with different modalities, for the diagnosis of deep infiltrating endometriosis [3–6].

Anorectal ultrasound scanning provides the most detailed view of endometriosis infiltration

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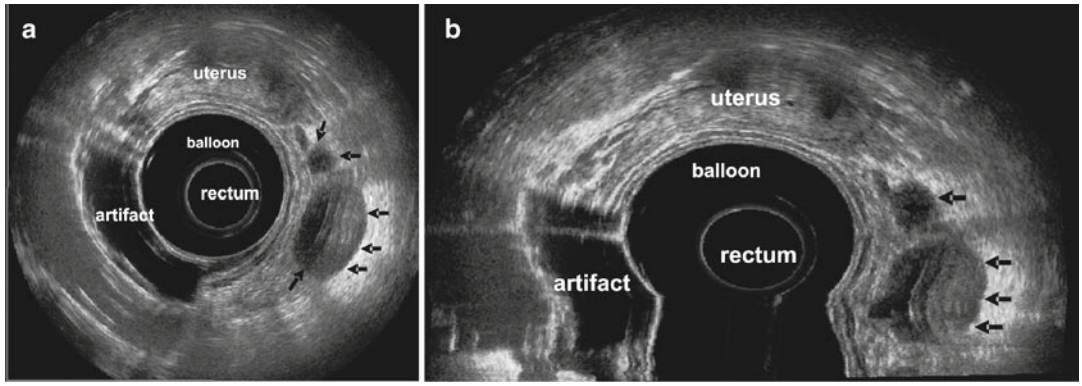

Fig. 10.1 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Endometriosis lesion infiltrating the perirectal fat. The rectal layers are intact. (**a**) Axial plane, (**b**) coronal with axial plane. Two heterogeneous hypoechoic images in the left lateral quadrant compromising the perirectal fat (*arrows*). Mucus in the rectal lumen, outside the lesion site (artifacts)

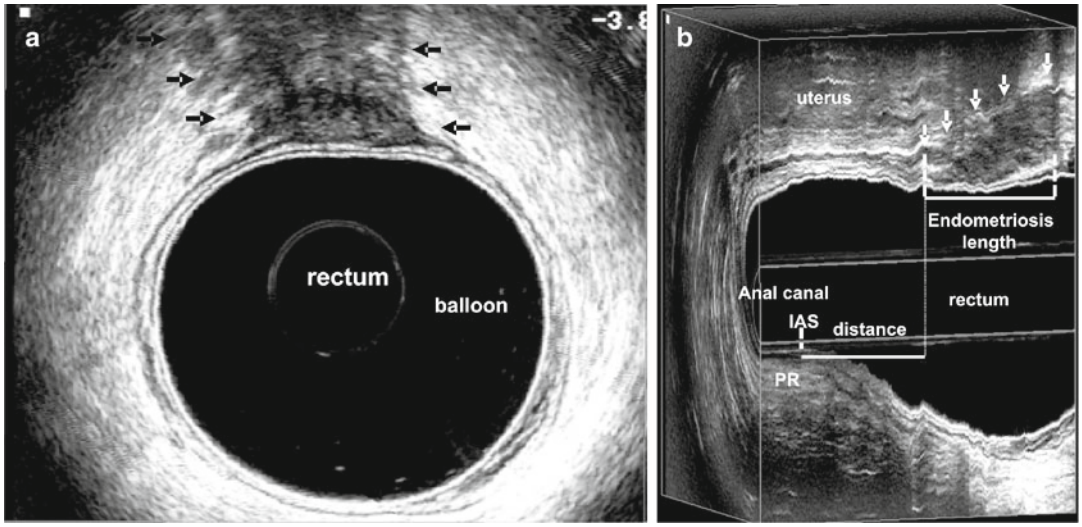

Fig. 10.2 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Endometriosis lesion in the anterior quadrant infiltrating the rectal wall as far as the muscular propria (*arrows*). (**a**) Axial plane. Heterogeneous hypoechoic image compromising 20 % of rectal circumference (*arrows*). (**b**) Sagittal plane. The length of the endometriosis lesion and the distance between the distal infiltration edge and the proximal edge of the sphincter muscles (posterior quadrant) (*arrows*). *IAS* internal anal sphincter, *PR* puborectalis muscle

in the rectum and mesorectal fat. The three-dimensional mode makes it possible to determine the exact circumferential and longitudinal extension of the infiltration into rectal wall or adjacent tissues and the distance between the distal infiltration edge and the proximal edge of the sphincter anal muscles [7], thus providing crucial information for the choice of therapeutic approach. Lesions appear as heterogeneous hypoechoic images mostly located in the recto-vaginal septum, in the mesorectal fat or serosa, and infiltrating into the muscular propria or submucosa layers (Figs. 10.1a, b, 10.2a, b, and 10.3a, b).

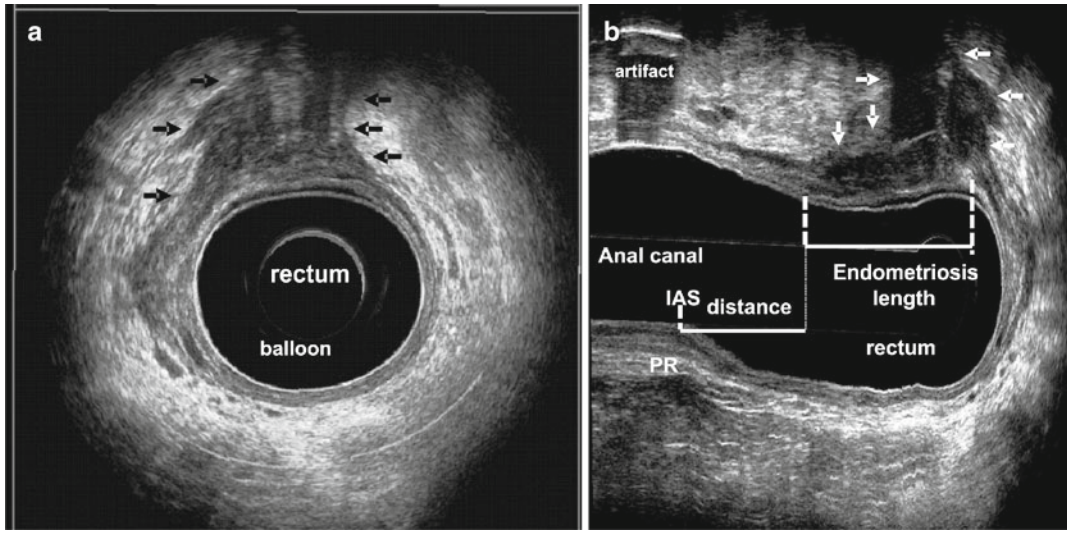

Fig. 10.3 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Endometriosis lesion in the right anterior quadrant infiltrating the rectal wall as far as the muscular propria. (**a**) Axial plane. Heterogeneous hypoechoic image compromising 30 % of rectal circumference (*arrows*). (**b**) Sagittal plane. The length of the endometriosis lesion and the distance between the distal infiltration edge and the proximal edge of the sphincter muscles (posterior quadrant) (*arrows*). Mucus in the rectal lumen, outside the lesion site (artifacts). *IAS* internal anal sphincter, *PR* puborectalis muscle

10.3 Presacral Neoplasia

Perirectal neoplasia is most often located in the retrorectal space and may be of varied etiology. Half the cases are congenital and two thirds are cystic in nature [8, 9]. It tends to affect young female adults but is uncommon in infants. Teratoma is the most frequently observed form in pediatric patients and contains fat or calcifications in 50 % of cases [9, 10].

A wide variety of cystic lesions occur in the retrorectal space, and most are congenital. They are classified as epidermoid cysts, dermoid cysts, enteric cysts (tailgut cysts and cystic rectal duplication), and neurenteric cysts according to their origin and histopathologic features [11]. Anorectal ultrasound may show specific signs and characteristics of the lesion (anechoic area with circular or oval shape, regular margin, and with reinforcement of posterior wall) but the diagnosis remains histopathologic. Ultrasonographic imaging is useful in the evaluation of size, type of lesion (mixed cystic and solid components), and relation with the rectal wall and the sphincter muscles (Figs. 10.4a, b and 10.5a–c).

Perirectal neoplasia appears with different characteristics: as a unilocular or multilocular retrorectal lesion, sometimes a hypoechoic area (cystic) or as an area of mixed echogenicity/heterogeneous image, due to mucoid material or inflammatory debris or solid component, usually with regular outline and not adhering to the rectal wall. In large lesion, an anorectal displacement or stenosis may be visualized due to extrinsic compression. It is important to define the rectal wall invasion or a communication between the cyst and the anorectal lumen (Fig. 10.6a–c).

10.4 Rare Tumors

10.4.1 Rectal Leiomyoma and Leiomyosarcoma

Leiomyoma is a benign mesenchymal neoplasm that usually develops where smooth muscle is present. This lesion is rare, except in the esophagus

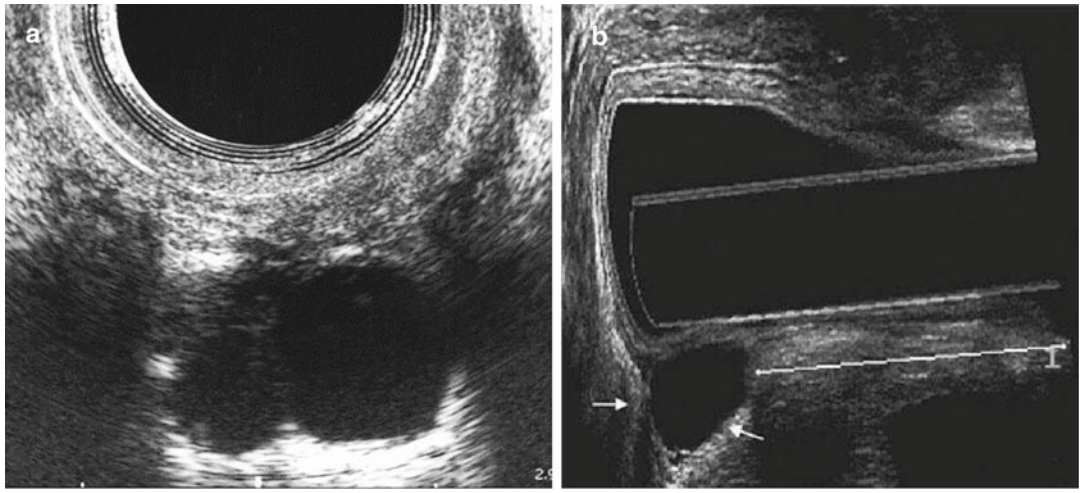

Fig. 10.4 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Female patient. Presacral developmental cyst. The lesion appears as a well-circumscribed, hypoechoic area with posterior reinforcement. The layers of the rectal wall are preserved. (**a**) Axial plane. (**b**) Sagittal plane. Lesion size (longitudinal length and the depth)

and rectum. Only 3 % of these smooth muscle tumors arising from colon are gastrointestinal leiomyomas and constitute about 0.1 % of rectal tumors [12, 13]. In the rectum, most leiomyomas present as small intraluminal polyps and are limited to the muscularis mucosa. Although there are reports of anorectal leiomyomas [14], definitive diagnosis requires anatomical and pathological examination (immunohistochemical staining). Leiomyomas are positive for actin and desmin and negative for CD34 and CD117 [13, 15].

Endorectal ultrasound scanning shows the exact extent of the lesion and relationship with the anatomical structures. Leiomyoma appears as a well-defined, homogeneous hypoechoic mass arising within and confined to the muscularis propria and without invasion of adjacent layers (Fig. 10.7a, b).

Leiomyosarcomas are malignant soft tissue neoplasms arising from smooth muscle tissue located within the muscularis propria and blood or lymphatic vessels. Histologically, leiomyosarcoma features spindle cells with elongated, blunt-ended nuclei in an eosinophilic cytoplasm. Immunohistochemically, these tumors are positive for vimentin, actin, smooth muscle myosin, and desmin. These lesions rarely metastasize through lymphatics and are more likely to spread through the lungs and liver through hematogenous spread.

10.4.2 Gastrointestinal Stromal Tumors

Gastrointestinal stromal tumors (GIST) are the most common mesenchymal tumors of the GI tract but they represent fewer than 1 % of all gastrointestinal (GI) tumors [16]. GIST can occur anywhere along the GI tract, but most often are found in the stomach (60 %) or small intestine (30 %), following rectum (3 %), colon (1–2 %), esophagus (<1 %), and omentum/mesentery (rare) [17].

The clinical presentation and diagnostic of patients with GIST depend on the anatomic location of the lesion and the size and aggressiveness. Small GIST may form solid subserosal, intramural, or less frequently, polypoid intraluminal masses. Large tumors tend to form external masses attached to the outer aspect of the gut involving the muscular layers [18]. The evaluation includes imaging and/or endoscopy but the

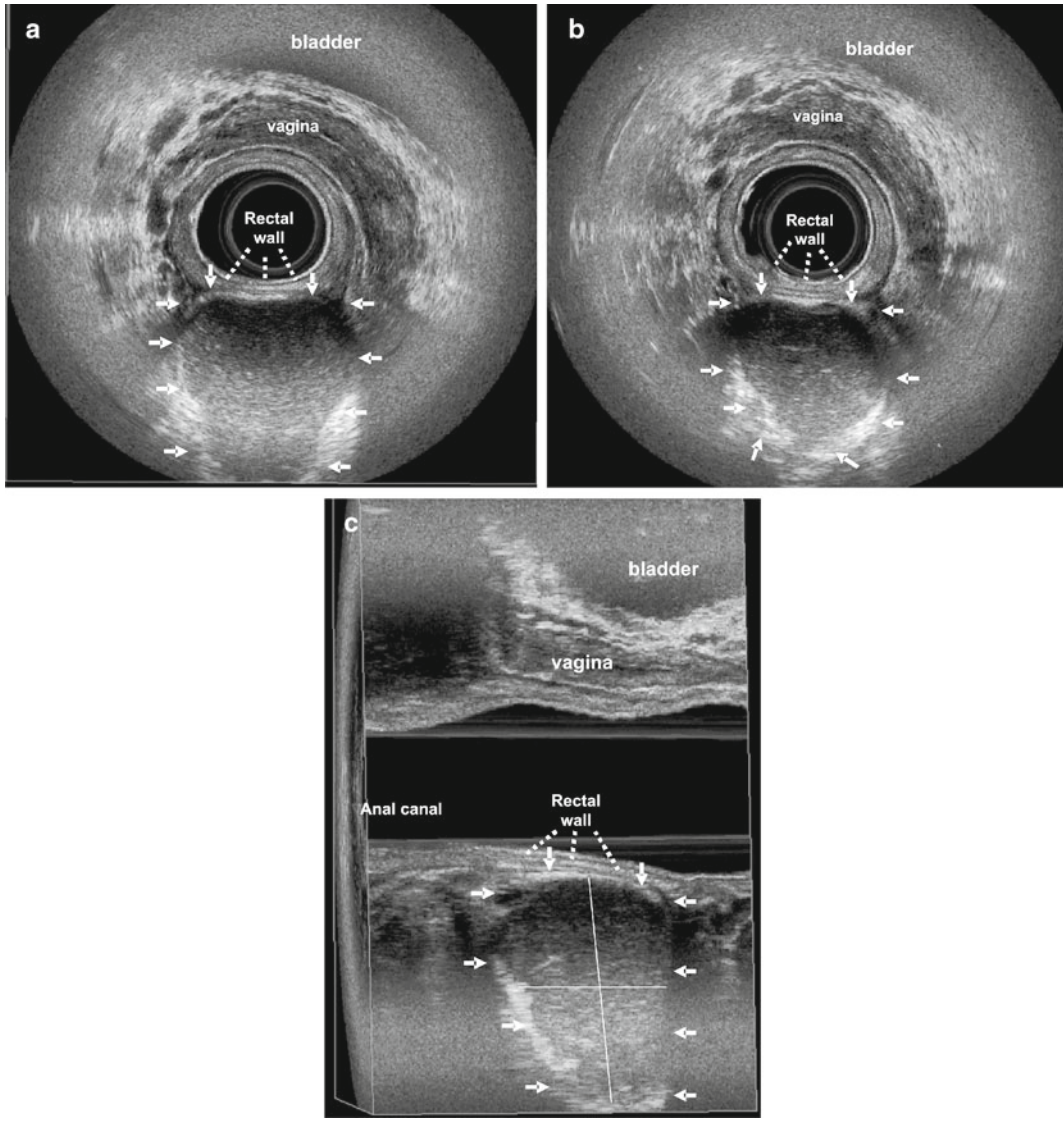

Fig. 10.5 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Female patient. Presacral cystic lesion located at the level of lower rectum with regular outline and without adherence to the rectal wall (*arrows*). The rectal wall is intact. (**a**, **b**) Axial plane. Mixed echogenicity lesion (*arrows*). (**c**) Sagittal with diagonal planes. A well-circumscribed (hyperechogenic line that surrounds the lesion) and unilocular cystic lesion. Lesion size (longitudinal length and the depth)

pathology and molecular genetics studies are required. Approximately 95 % of GISTs are positive for the CD117 antigen [18].

Anorectal ultrasound scanning provides the most detailed view of lesion and the relationship with the anatomical structures, including perirectal, perianal tissues, anal canal muscles, perirectal wall, and adjacent organs. On ERUS, GIST appears as a well-defined round, homogeneous hypoechoic mass arising from the muscularis propria with an overlying intact mucosa (Fig. 10.8a–c).

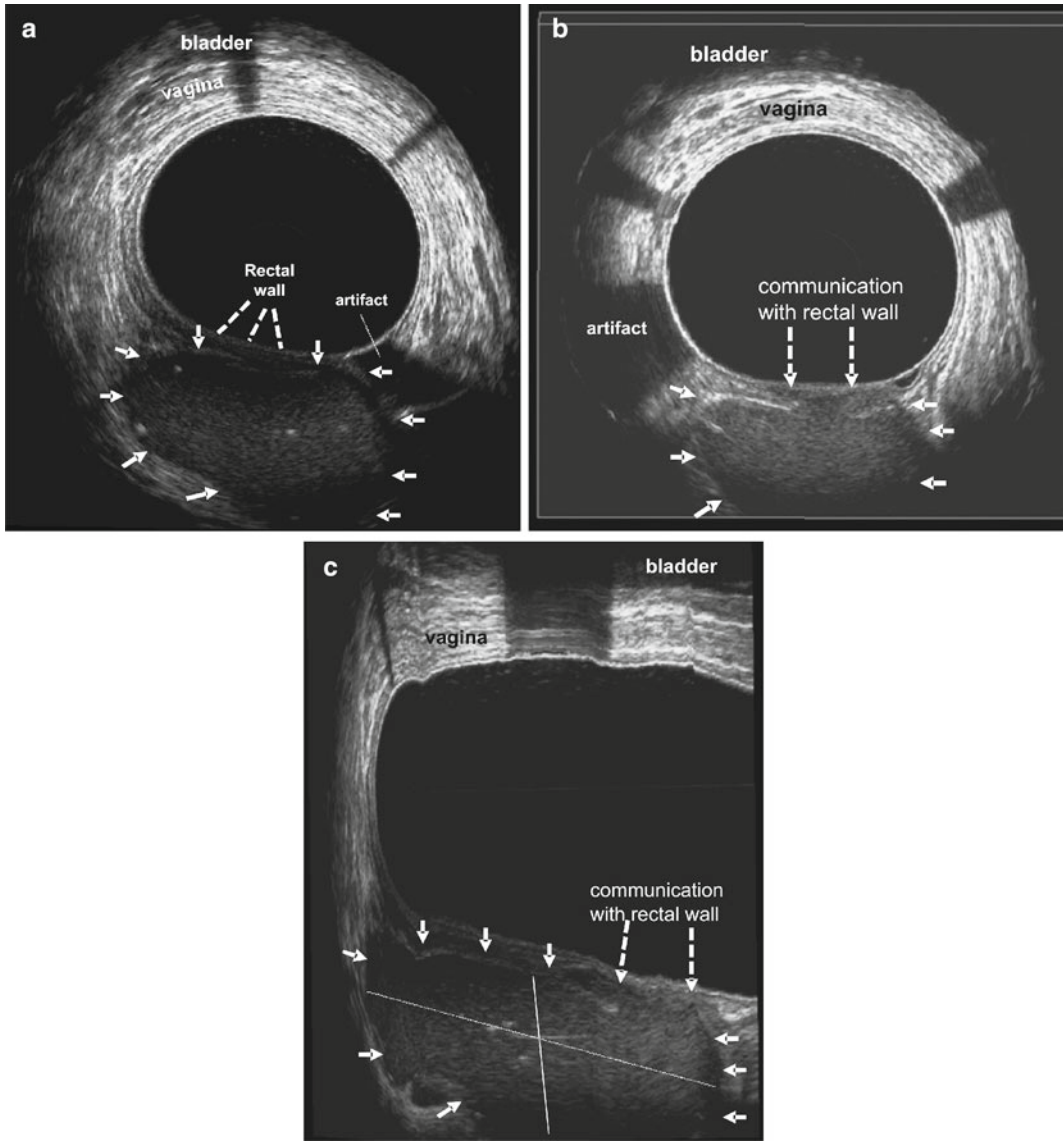

Fig. 10.6 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Female patient. Cystic lesion in the presacral space at the level of lower rectum. There is a contiguous (communication) area with rectal wall. (**a**) Axial plane. In this position, the lesion appears with mixed echogenicity, with regular outline and without adherence to the rectal wall (*arrows*). The rectal wall is intact. (**b**) Axial plane. The image shows the area of the cystic lesion, which communicates with the rectal wall (*interrupted arrows*). (**c**) Sagittal plane. The hyperechogenic line that surrounds the lesion (*arrows*) is interrupted (small area) and there is a communication with rectal wall (*interrupted arrows*). Lesion size (longitudinal length and the depth)

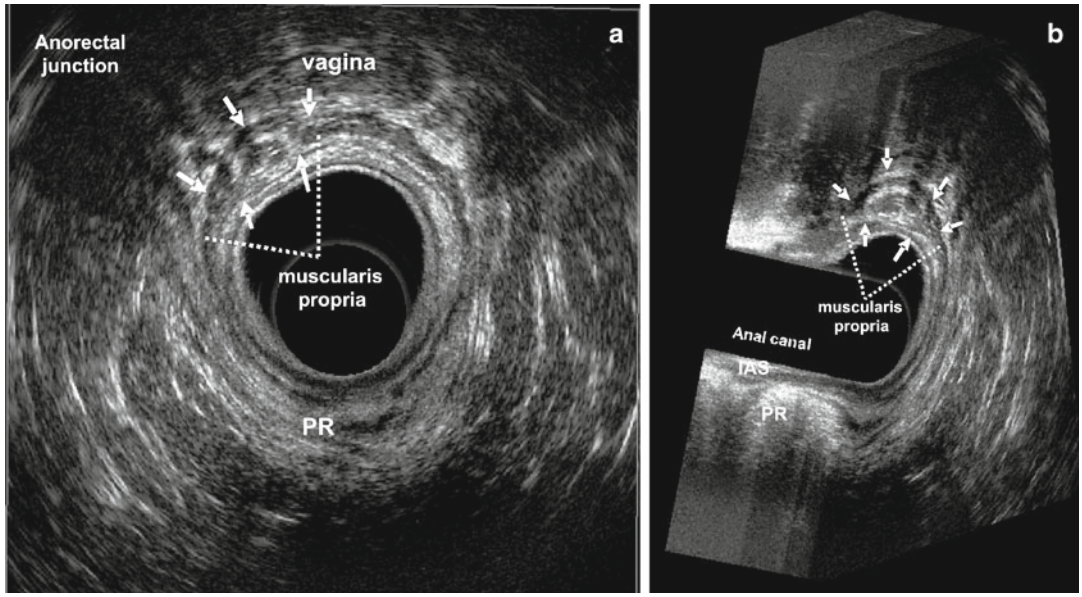

Fig. 10.7 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Small leiomyoma located at the level of the anorectal junction in the right anterior quadrant. The mixed echogenicity lesion expands the outer hypoechoic layer that corresponds to the muscularis propria (*arrows*). (**a**) Axial plane; (**b**) multiplanar: sagittal with diagonal and axial planes. *IAS* internal anal sphincter, *PR* puborectalis muscle

10.4.3 Angiosarcoma

Angiosarcoma accounts for 4 % of all soft tissue malignancies. Tumor originates from vascular and lymphatic walls. It presents as firm, highly vascular lesions. Immunohistochemical stains are often employed to confirm the diagnosis and they include CD 31, CD 34, and BNH 9 (an endothelial marker).

Endorectal ultrasound can demonstrate the presence of vascular structures, appearing as anechoic areas within mixed echogenicity lesion (Fig. 10.9a, b).

10.4.4 Rhabdomyosarcoma

Rhabdomyosarcoma is one of the most common childhood soft tissue tumors, but represents less than 5 % of malignant soft tissue lesions in adult. Anorectal presentation is extremely rare and is seen less than 2 % of cases. It arises from the muscular layer of the bowel. It is described as a grossly uncircumscribed lesion with multiple areas of spherical growth, often resembling a "bunch of grapes" that is soft in consistency. As a result of its mesodermal origin, rhabdomyosarcoma tends to show multiple areas of muscle tissue origin at different stages of development.

This tumor is harder to diagnose in adults, with more advanced disease at presentation and worse prognosis than the younger age groups.

10.4.5 Schwannoma

Schwannoma arises from neural crest cells and can therefore occur in any anatomical region. Grossly these tumors often appear as firm yellow- or brown-colored lesions, which may be pedunculated or sessile. They are almost always restricted to the submucosa and have largely benign slow-growing nature. They are uniformly S-100 positive.

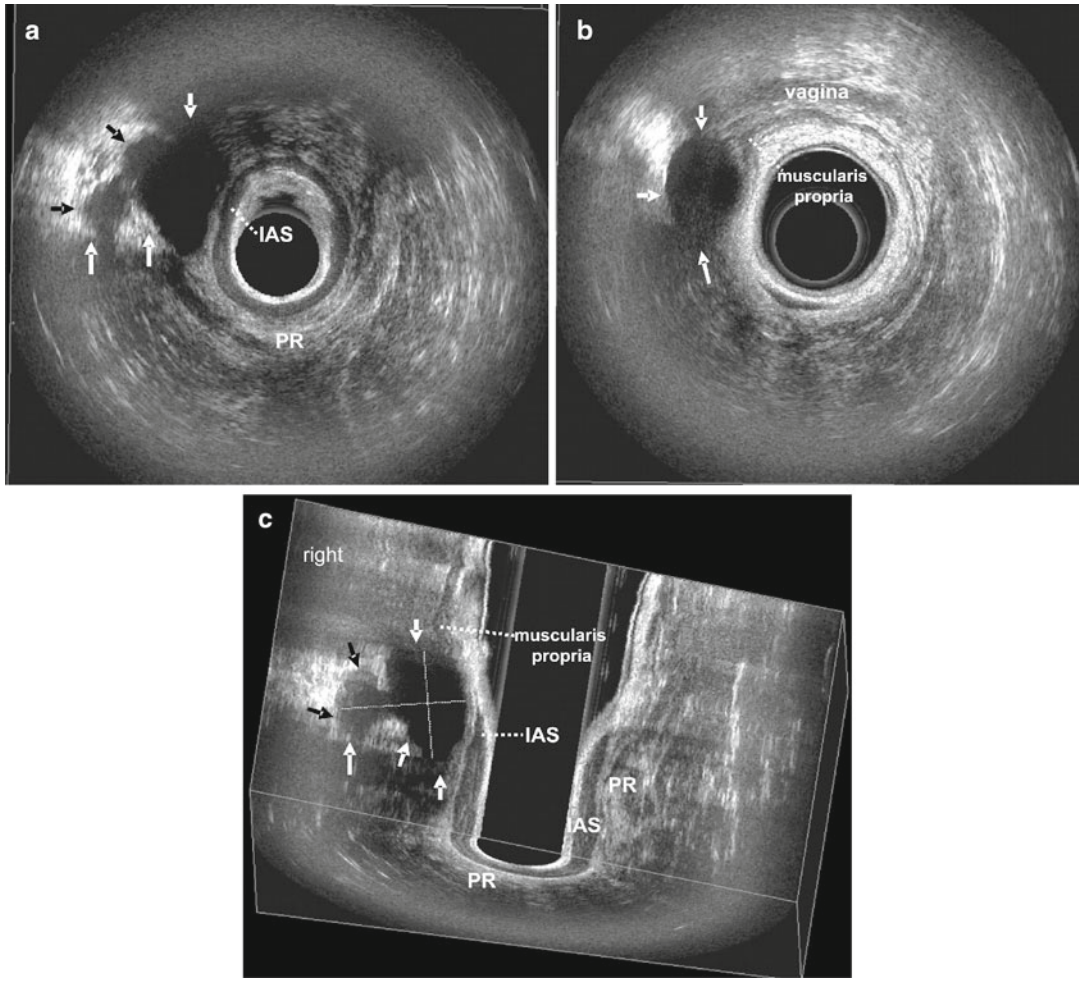

Fig. 10.8 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Female patient. Mixed echogenicity lesion located at the level of lower rectum, anorectal junction, and upper anal canal in the right anterior quadrant. The lesion involved the puborectalis muscle (PR) and the rectal wall. (**a**) Axial plane. Lesion located in the perianal and ischiorectal fat and involving the PR muscle (right anterior quadrant) (*arrows*). The internal anal sphincter (IAS) is intact; (**b**) axial plane. The lesion is located in the perirectal fat and involves the rectal wall as far as the muscular propria (*arrows*); (**c**) coronal plane. Lesion size (longitudinal length and the depth) (*arrows*)

10.5 Conclusions with Future Research

Anorectal cysts, endometriosis of the rectovaginal septum, and non-mucosal rectal lesions (rare neoplasias of the muscularis propria, connective tissues, and vascular structures) are well visualized with 3D endoanal ultrasonography. The lesions are easily available for ultrasound examination to set the course for surgical or non-surgical management. The utilization of this technology largely depends on the availability of the technology to the surgeon.

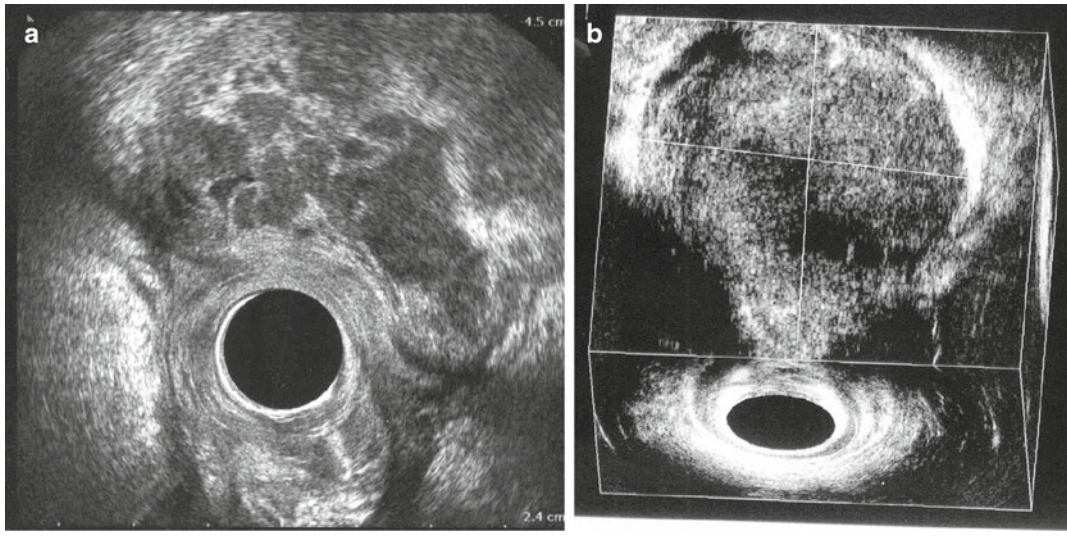

Fig. 10.9 Three-dimensional endorectal ultrasound performed with 2050 transducer (B-K Medical). Female patient. Large mixed echogenicity angiosarcoma with anechoic area inside (vascular structures) located at the level of lower rectum in the anterior quadrant. The lesion is located in the perirectal fat and involves the rectal wall as far as the muscular propria. (**a**) Axial plane; (**b**) coronal plane

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