Diagnosis of deep endometriosis: clinical examination, ultrasonography, magnetic resonance imaging, and other techniques

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The aim of the present review was to evaluate the contribution of clinical examination and imaging techniques, mainly transvaginal sonography and magnetic resonance imaging (MRI) to diagnose deep infiltrating (DE) locations using prisma statement recommendations. Clinical examination has a relative low sensitivity and specificity to diagnose DE. Independently of DE locations, for all transvaginal sonography techniques a pooled sensitivity and specificity of 79% and 94% are observed approaching criteria for a triage test. Whatever the protocol and MRI devices, the pooled sensitivity and specificity for pelvic endometriosis diagnosis were 94% and 77%, respectively. For rectosigmoid endometriosis, pooled sensitivity and specificity of MRI were 92% and 96%, respectively fulfilling criteria of replacement test. In conclusion, advances in imaging techniques offer high sensitivity and specificity to diagnose DE with at least triage value and for rectosigmoid endometriosis replacement value imposing a revision of the concept of laparoscopy as the gold standard. (Fertil Steril® 2017;108:886–94. ©2017 by American Society for Reproductive Medicine.)

Key Words: Endometriosis, deep endometriosis, clinical examination, transvaginal sonography, MR imaging

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Pelvic endometriosis is defined as the presence of endometrial tissue outside the endometrium and myometrium (1). Overall it is estimated to affect around 10% of women of reproductive age, increasing to 35%–50% in symptomatic patients (2, 3). Three main entities of pelvis endometriosis have been identified: peritoneal, ovarian or deep endometriosis (DE) (4).

DE is thought to affect 20% of women with pelvic endometriosis and is a source of pain and infertility (5, 6). However, while the three forms are often associated, in contrast to peritoneal and ovarian endometriosis, no clear consensus exists on the definition of DE. Based on the relation between the depth of infiltration and intensity of pain, it has been arbitrarily defined as endometriosis infiltrating the peritoneum by $>$ 5 mm (7). The same authors have recently suggested that DE should be pathologically defined as adenomyosis externa (8). However, in accordance with a recent Cochrane metaanalysis, DE is defined in the present review as a fibrous/muscular infiltration of organs and anatomical structures containing endometrial tissue below the peritoneum, regardless the depth of infiltration (9).

Independently of the issue of DE definition, symptomatic patients with or without suggestive clinical examination, require additional routine investigations mainly comprising transvaginal sonography (TVS) and MR imaging (MRI), to determine therapeutic strategy. The goals of this review are to analyze the accuracy of clinical examination and imaging techniques to assess DE locations, to evaluate whether imaging techniques may replace the gold standard of diagnostic laparoscopy for some locations of DE, and finally, to determine

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whether imaging DE mapping could have implications on surgical management.

CLINICAL EXAMINATION

The first step in diagnosing DE is to establish the patient's clinical history with particular emphasis on symptoms (dysmenorrhea, dyspareunia, dysuria, dyschezia, and chronic pelvic pain) as well as, age, height, weight, ethnic origin, gravidity, parity, previous surgery for endometriosis, family history of endometriosis, previous non-surgical treatment for endometriosis, and infertility. However, several authors have underlined the poor relationship between symptoms exhibited by patients and the severity of the lesions rendering clinical diagnosis difficult (10–12). Moreover, it is thought that 2% to 50% of women could have asymptomatic endometriosis (10–12).

The second step is based on physical examination including a systematic analysis of the posterior vaginal fornix with a speculum to look for retraction and dark nodules. Digital examinations should be performed of the vagina to assess the characteristics of the uterus and adnexa, of the vesico-uterine pouch to detect bladder invasion, and of the retrocervical area to detect infiltration of the torus uterinum, uterosacral ligaments (USLs), pouch of Douglas (POD), vagina, and rectovaginal septum (RVS). Rectal digital examination can help in assessing the involvement of the rectum, parametrium and visceral pelvic fascia.

In the particular setting of DE, few data are available to evaluate the accuracy of physical examination. One retrospective study found that routine clinical examination detected DE in only 36% of 140 women with DE, and the authors suggest the accuracy of physical examination improves during menstruation (13). To detect rectosigmoid and retrocervical DE without differentiating between the different specific DE locations, Abrao et al. (14) reported that digital vaginal examination had a sensitivity of 72% and 68%, a specificity of 54% and 46%, a positive predictive value (PPV) of 63% and 45%, and negative predictive value (NPV) of 64% and 69%, respectively.

In our experience, even when the examination is performed by an expert, the sensitivity, positive (PLR) and negative (NLR) likelihood ratios are 73.5%, 3.3 and 0.34 for uterosacral ligament endometriosis, 50%, 3.88, and 0.57 for vaginal endometriosis, and 46%, 1.67, and 0.75 for intestinal endometriosis, illustrating the limits of physical examination (15). Moreover, clinical examination is further complicated by the high prevalence of myofascial trigger points in the pelvic floor in women with DE, a source of severe pain limiting the evaluation of DE locations (16).

ULTRASOUND

A recent international consensus highlighted the need for a reliable diagnostic system of triage to evaluate the location and the extent of DE (17). In this setting TVS emerges as the first-line imaging technique due to its availability and relatively low cost. In addition to the sonographic description of DE lesions, the operator should explore the peritoneum for superficial implants, the uterus for adenomyosis, and the ovaries for endometriomas. A transabdominal scan of the kidneys should also be systematic to detect hydronephrosis. This detection is important, as diagnosis of ureteral involvement in pelvic area is often difficult using imaging techniques. Moreover, hydronephrosis can be asymptomatic and can compromise the kidney function requiring surgical management with at least ureteral stent.

Standardized, consensual terminology describing TVS appearance and anatomical locations is essential in the diagnosis of DE (17). Lesions appear as hypo- or isoechoic solid nodules, which may vary in size and have smooth or irregular contours, or as hypoechoic thickening of the wall of bowel, vagina, and bladder (18, 19).

The distribution of DE nodules should be evaluated in the whole pelvic cavity including the anterior, posterior, and subperitoneal lateral compartments. In accordance with previous studies, DE lesions are most frequently located in the posterior compartment, involving the torus, USLs, vagina, RVS, POD, and rectosigmoid colon (6, 20). Less frequently, anterior DE locations are present involving the vesico-uterine pouch, bladder, and round ligaments. Finally, rarely described by TVS, lateral compartment involvement includes the parametrium, ureter, visceral fascia, and lateral pelvic wall. The accuracy of sonography should be analyzed according to the DE locations and the specific sonographic techniques used.

Guerriero et al. (21) performed a preliminary comparison between 'tenderness-guided' TVS and 3D-TVS to detect pelvic endometriosis independently of location, and reported that 'tenderness-guided' TVS was less accurate. Sonovaginography (SVG) is the combination of TVS with the introduction of a gel or saline solution into the vagina creating an acoustic window between the transvaginal probe and the surrounding structures of the vagina (22). Dessole et al. (22), comparing SVG to TVS, showed that SVG had higher sensitivity and specificity. Finally, independently of the DE location, Nisenblat et al. reported a pooled sensitivity and specificity for all TVS techniques (TVS, 3D-TVS, and SVG) of 79% and 94%, which approach the criteria for a triage test (9).

TVS Evaluation of the Posterior Pelvic Compartment

The torus uterinum is not clearly defined and has rarely been mentioned in previous TVS reports due to difficulties in evaluating the posterior wall of the uterus, particularly for retroversed and retroflexed uteri (23). In some cases, a nodular hypoechoic thickening located just behind the cervix above the posterior vaginal fornix may be suggestive of a diagnosis of DE (Fig. 1) (23).

Although normal USLs are usually not visible on ultrasound (18), they can sometimes appear as a thin regular lateral hyperechoic strand in the presence of pelvic fluid in the POD (23). A USL is considered to be involved by DE when a lateral, echoic, regular or irregular linear thickening is visible in the subperitoneal fat, mainly behind the upper part of vagina (Fig. 2) (17, 19, 23). Two recent meta-analyses of USL endometriosis have reported pooled sensitivities and specificities of 53%–64% and 93%–97%, respectively (9, 24). The contribution of rectal endoscopic sonography (RES) for USL endometriosis was only evaluated by one study and