EndoShare: Focus on Endometriosis Diagnosis

Introduction

Endometriosis is defined as a chronic inflammatory disease characterized by the presence of endometrial tissue outside the uterus that causes chronic pelvic pain, dysmenorrhea, dyspareunia, and infertility,[1] all of which result in significant morbidity and diminished patient quality of life.[2] Data estimate that up to 10% of women of reproductive age may suffer from endometriosis.[3,4] But since many females with endometriosis are undiagnosed,[5] a significant proportion of women experience the symptoms of endometriosis without understanding the cause or their appropriate management.[6]

Typical symptoms of endometriosis include dysmenorrhea, dyspareunia, menorrhagia, non-menstrual cycle pelvic pain, dysuria, chronic fatigue, and infertility.[7,8] These symptoms can disrupt work and home lives and can contribute to physical and emotional suffering.[9-11] Although diagnosis of endometriosis may occur in the second or third decade of life, one-third of patients first experience symptoms before the age of 15; the average delay in diagnosis is >9 years.[12] Several factors contribute to this diagnostic delay, including misdiagnosis and "normalization" of symptoms by the patient.[13]Of note, diagnostic delay in endometriosis is a worldwide problem.[13] That is why an early and accurate diagnosis is critical to the optimal management of the disease. Indeed, failure of timely diagnosis and adequate treatment may lead to disease progression compromising fertility and increasing the risk of chronic pelvic pain.[14-16]

Differential Diagnosis

Chronic pelvic pain may include endometriosis but may also include adenomyosis, pelvic inflammatory disease, or ovarian or tubal masses.[17] Moreover, pelvic pain may not necessarily be due to gynecologic causes and may be due to gastrointestinal, urinary, neurologic, and musculoskeletal disorders.[17] The presence of endometriotic lesions does not preclude other etiologies accounting for the patient's symptoms, but the lack of obvious lesions does not preclude the possibility of endometriosis.[6] Further complicating its diagnosis is the poor correlation between symptoms as well as the severity and extent of the disease.[18]

Gastrointestinal etiologies of non-menstrual pelvic pain include:[19]

- Irritable bowel syndrome (IBS)
- Inflammatory bowel disease (IBD)
- Celiac disease
- Chronic constipation
- Diverticular disease
- Cancer (colon) – in the older patient

Urologic etiologies of non-menstrual pelvic pain include:[19]

- Interstitial cystitis (the "evil twin" to endometriosis)[20]
- Recurrent urinary tract infection (UTI)

- Stone(s) in the bladder
- Urethral syndrome
- Pelvic floor dysfunction

Differentiating endometriosis from these conditions may be difficult as symptoms are similar and may follow a cyclic or constant pattern.[17] A thorough examination to exclude other causes of pelvic pain should be conducted before instituting aggressive therapy for endometriosis. Many clinical practice guidelines for endometriosis recommend the treatment of symptoms before obtaining a definitive surgical diagnosis.[17,21,22] Although biomarkers have been proposed, they are not recommended for the diagnosis of endometriosis, as none have been validated for endometriosis.[23]

"Endometriosis is a very different disease than arthritis of the hand or elbow. Endometriosis affects the whole family—the whole family has pain. You can't help with homework. You can't go to the soccer game. Your whole life is affected. As a physician, you need to take a more global viewpoint—beyond simply the patient".

- Stephen M. Cohen, MD, FACOG

Patient Interviews – Asking the Right Questions

Diagnosis should be based on a thorough process of patient interviews, clinical examination, and imaging;[1] guidelines state that exploratory laparoscopy is no longer necessary to make a presumptive diagnosis of endometriosis prior to beginning treatment.[Error! Bookmark not defined.] Questions that may be asked during patient interviews include:[19]

- What is the pain like?
- Where is the pain?
- When did the pain start?
- What makes the pain worse?
- What makes the pain better?
- Is the pain cyclic?
- Is there any effect from food, intercourse, bowel movements, or urination?
- What else may be associated with the pain?

"Just listen to your patient; she [he] is telling you the diagnosis."

-Adapted from Sir William Osler (1849-1919)

Assessing the level of pain in patients with symptoms of endometriosis can be difficult for physicians.[17] Methods of pain assessment include the visual analog scale, the McGill questionnaire, and quality of life scales, such as the SF-36 (see Table for additional assessment tools). Such methods can also be used to assess treatment response to therapy.[17]

Table. Various Pain Assessment Tools for Endometriosis.[24]

Assessment Tool	Features	Limitations
Visual Analog Scale (VAS)	Ranges from "no pain" to "worst" for any pain with endometriosis; validated; easy to administer; reliable	Subjective pain measure
Numerical Rating Scale (NRS)	Ranges from 0-10 for any pain with endometriosis; validated; easy to administer; reliable	Subjective pain measure
Biberoglu & Behrman (B&B)	Specific to endometriosis; multidimensional (dysmenorrhea, pelvic pain, dyspareunia)	Subjective pain measure; only HCP can administer
McGill Pain Questionnaire	Validated; multidimensional (pain location, intensity, quality, pattern, alleviating/aggravating factors); reliable	Subjective pain measure; only HCP can administer
Andresch & Milsom's scale	Numerical scale (0-7), multidimensional	Subjective pain measure

Current evidence may support the clinical diagnosis of endometriosis as opposed to a surgical diagnosis;[6] this path may also potentially reduce diagnostic delay. Clinical assessments that may be used to diagnose endometriosis include, but are not limited to:[6,25]

- **Symptoms**: pelvic pain that is chronic, cyclic, and persistent or progressive; dysmenorrhea; non-menstrual pelvic pain; dyspareunia; dysuria
- **Patient and family history**: history of infertility; previous pelvic surgery; history of benign ovarian cysts and/or ovarian pain
- **Menstrual cycle characteristics**: heavy menstrual bleeding; excessive/irregular bleeding; passing clots; irregular menstrual periods
- **Pelvic/physical examination**: abdominal palpitation done slowly and with care, proceeding to bimanual pelvic examination; note anatomical location, palpitation of the uterine/bladder pouch, the Douglas pouch and adnexa can reveal painful sites; always note patient's face during the examination as it will show areas of more intense pain
- **Combination of assessments**: combining number of symptoms with history of the patient and ultrasound findings
- **Additional considerations**: imaging; transvaginal ultrasound in conjunction with symptoms, patient history, and/or physical findings; ultrasound is particularly sensitive for detecting ovarian endometriomas as well as deep endometriosis

Detecting endometriosis through laparoscopy relies on the visualization of lesions, which are described as a "classic powder burn, red and clear lesion, ovarian disease, and thickening of the uterosacral ligament."[19] Unfortunately, visualization may be challenging due to a heterogeneous lesion appearance, lesion location that is inaccessible (e.g., deep), and interobserver variability.[6] Furthermore, the positive predictive value of visualization via laparoscopy ranges from 43% to 45%.[26,27]

Conclusions

Clinical practice guidelines for the treatment of endometriosis and pain associated with endometriosis suggest that endometriosis is best viewed as a chronic medical disease requiring lifelong management through the optimal use of medical treatment and avoidance of repeated surgical procedures (e.g., laparoscopic ablation, excision, etc.). Thus, endometriosis is associated with significant morbidity. Persistent pelvic pain, especially in the presence of other symptoms associated with endometriosis, patient history, and results of a physical examination suggest endometriosis. Transvaginal ultrasound and laparoscopy may be beneficial when findings are unclear. And perhaps most importantly, an accurate and timely diagnosis of endometriosis leads to effective early treatment.

"I think the more you listen, the more the patient understands that you're concerned. You may not completely eliminate the pain, but they understand you want to help them the best you can. Set realistic goals. When we work together with the patient, we can offer effective, compassionate care.

- Stephen M. Cohen, MD, FACOG

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