

Lown Right Care

Reducing Overuse and Underuse

Avoiding Delays in Diagnosing Endometriosis

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Patient perspective by Helen Haskell and John James

Case Scenario

A 27-year-old woman presents for a new patient visit to discuss chronic, severe menstrual-related pain that began at menarche when she was 12 years of age, causing her to miss many days of school and work. Over the past few years, she has been examined by her primary care physician several times and consulted a gynecologist for the pain, which is associated with nausea, diarrhea, and abdominal bloating. Results of abdominal and pelvic examinations and laboratory testing, including a complete blood count, comprehensive metabolic profile, thyroid-stimulating hormone level, progesterone level, and tests for sexually transmitted infections, were normal. Dysmenorrhea was diagnosed, and the patient was treated with ibuprofen, naproxen, and several trials of oral contraceptives, with little or no relief. She is constantly tired and feels anxious and depressed. She is struggling at work, especially when she has her menstrual period, and recently broke up with her significant other because of her labile moods and emotional outbursts. She is tearful and appears to be desperate for help.

Clinical Commentary

EPIDEMIOLOGY

Endometriosis is an idiopathic inflammatory, estrogen-dependent condition caused by the presence of endometrial tissue outside of the uterus.¹ Endometriosis is characterized by painful and disabling menstrual symptoms. The most commonly reported symptoms are painful periods (62%), heavy/irregular bleeding (51%), and pelvic pain (37%).²

Endometriosis affects approximately 10% of reproductive-aged women and others assigned female sex at birth.³ Endometriosis is more common in people with onset of menarche before 12 years of age, menstrual cycles of less than 24 days, lower parity, and lower lean body weight (i.e., the difference between total body weight and body fat weight).³ Up to 50% of people with concomitant infertility and chronic pelvic pain have endometriosis.³ Endometriosis has been called a “missed disease” due to its unclear etiology, lack of research, and inconsistency in diagnosis and management, leading to frequent diagnostic and therapeutic delays.⁴

Multiple studies have confirmed the significant economic burden of endometriosis, with associated medical costs totaling \$78 to \$119 billion annually in the United States.⁵ Significantly higher costs are incurred in patients who experience longer diagnostic delays.⁵⁻⁷ The effect of endometriosis on home or work productivity and absenteeism can be substantial, with the average patient losing more than 10 hours of weekly productivity.^{8,9}

Endometriosis is associated with a significantly lower quality of life, depression, increased self-harm, and the development of multiple medical comorbidities, including autoimmune disease, inflammatory bowel disorders, and psychiatric disorders.¹⁰⁻¹² Endometriosis is also linked to a higher risk of ovarian cancer.¹⁰⁻¹² The lack of a clear path to a diagnosis often leads clinicians and patients to inappropriately dismiss symptoms as a normal part of the menstrual cycle.¹³ Patients are also often reluctant to disclose menstrual symptoms.¹⁴ The stigma of endometriosis is

Lown Institute Right Care Alliance is a grassroots coalition of clinicians, patients, and community members organizing to make health care institutions accountable to communities and to put patients, not profits, at the heart of health care.

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CME This clinical content conforms to AAFP criteria for CME. See CME Quiz on page 205.

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TAKE-HOME MESSAGES FOR RIGHT CARE

Endometriosis is a common and debilitating condition associated with significant morbidity, stigmatization, and decreased quality of life.

To avoid delays in diagnosis, a methodical and comprehensive approach to the evaluation of patients with suspected endometriosis is essential.

Nonsteroidal anti-inflammatory drugs, hormone therapy, and complementary therapies should precede any surgical intervention.

associated with adverse effects on physical and mental health, deterioration of family and social interactions, and dismissive attitudes of health care professionals.¹⁵

CLINICAL PRESENTATION AND BENEFITS OF EARLY DIAGNOSIS

Patients with endometriosis commonly present with severe menstrual pain, cramping, and heavy menstrual bleeding. Other symptoms include dyspareunia, fatigue, dysuria, rectal pain, diarrhea, and infertility.¹⁶ No laboratory tests are helpful in the diagnosis of endometriosis, but tumor markers in women older than 50 years may help exclude malignant conditions.^{17,18} The clinical presentation of endometriosis can be confused with many gynecologic, gastroenterologic, and mental health conditions, leading to an average diagnostic delay of four to 11 years and subsequent treatment delays.¹⁹⁻²¹ The average patient sees seven physicians before endometriosis is diagnosed.²²

Although laparoscopic biopsy is considered the definitive method of diagnosis, recent guidelines recommend offering empiric treatment if there is a high suspicion of endometriosis based on clinical assessment and transvaginal ultrasonography or pelvic magnetic resonance imaging.²³ Nonsteroidal anti-inflammatory drugs and hormone therapy (combined oral contraceptives or progestins alone) are first-line treatments. Adjunct therapies include an anti-inflammatory diet, which consists of fruits and vegetables, whole grains, lean protein, healthy fats, and spices, and limits processed foods, red meats, alcohol, gluten, and dairy. Other complementary modalities include exercise, acupuncture, and nutritional supplements.^{15,16,19} Surgery should be considered only when symptoms are persistent and severe enough that the benefits outweigh the risks.²⁴ A patient-centered, shared decision-making discussion and multiple failed therapeutic trials should precede discussion of surgery.¹⁶

Endometriosis is associated with diagnostic delays due to its nonspecific symptoms, lack of specific tests, and a lack of awareness by patients and physicians.^{16,25,26} These delays can be mitigated by obtaining a meticulous history and following a comprehensive, patient-centered approach to care. It is essential to avoid diagnostic overshadowing by dismissing symptoms as functional or psychosomatic, or normalizing them as a painful menstrual period.^{25,27} Although it has not

been established that early diagnosis of endometriosis alters the course of the disease, avoiding delays in diagnosis can mitigate significant distress and impaired quality of life.²⁶ The natural course of endometriosis is periods of remissions and exacerbations with eventual resolution at menopause.¹⁶

Patient Perspective

Because of the widespread delays in diagnosis and the morbidity caused by endometriosis, more attention to a timely diagnosis and treatment strategies is needed. Endometriosis is a hot-button issue in women's pain. It is one of a suite of women's health issues that reformers charge are not taken seriously by doctors; the term gaslighting, now freely mentioned in critiques of women's health care, refers as often to endometriosis as to any other condition.²⁸ Varying reports on the effectiveness of the few treatments available do not help the tension in the doctor-patient relationship created by the difficulty in determining a diagnosis.²⁹

Recent research findings about the causes of endometriosis could lead to new treatment options.³⁰⁻³² Until then, the unenviable job of the family physician is to navigate the uneven terrain of evolving science, patient experience of illness, and the limitations of existing treatments. In this context, the most important things the physician can provide are up-to-date information, a thorough and sympathetic diagnostic process, referral to specialists when needed, and moral support and guidance in what is often a difficult journey. Joining an endometriosis support group may help the patient feel less alone in dealing with the ups and downs of this disease.

Resolution of Case

The patient's new physician recognized her history and classic symptom presentation as a potential case of endometriosis. Subsequent transvaginal ultrasonography followed by pelvic magnetic resonance imaging confirmed the likely diagnosis of endometriosis without the need for laparoscopy. The patient was relieved to have a diagnosis and felt supported by her physician. After a comprehensive patient-centered discussion of the natural course of the disease and treatment options, she resumed a regimen of nonsteroidal anti-inflammatory drugs and hormone therapy, initiated new lifestyle changes, mindfulness activities, and complementary therapies, and began feeling much better.

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