

ENDOMETRIOSIS: MODERN PERSPECTIVES ON PATHOGENESIS, DIAGNOSIS, AND TREATMENT

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Received: 2026-04-10 · Accepted: 2026-05-25

Abstract

Endometriosis is a chronic estrogen-dependent gynecological disorder characterized by the presence of endometrial-like tissue outside the uterine cavity. It affects millions of women worldwide and is one of the leading causes of chronic pelvic pain, infertility, and reduced quality of life. Despite extensive research, the exact mechanisms underlying the development of endometriosis remain incompletely understood. Recent advances in molecular biology, genetics, immunology, and imaging technologies have significantly improved understanding of disease pathogenesis and expanded opportunities for early diagnosis and individualized treatment. The aim of this study was to analyze contemporary concepts regarding the pathogenesis, diagnostic approaches, and therapeutic strategies for endometriosis. The findings indicate that endometriosis is a multifactorial disease involving hormonal, immunological, inflammatory, genetic, and environmental factors. Early recognition and comprehensive management can substantially improve clinical outcomes and reproductive health. Modern treatment strategies combining medical therapy, minimally invasive surgery, and multidisciplinary care contribute to symptom control and improved quality of life.

Keywords: endometriosis, chronic pelvic pain, infertility, estrogen, inflammation, laparoscopy, reproductive health, hormonal therapy, gynecology, women's health.

1. Introduction

Endometriosis is recognized as one of the most complex and challenging disorders in modern gynecology. The disease is characterized by the growth of endometrial-like glands and stromal tissue outside the uterine cavity, most commonly affecting the ovaries, pelvic peritoneum, uterosacral ligaments, rectovaginal septum, and surrounding pelvic structures. In severe cases, lesions may also involve the urinary tract, intestines, diaphragm, and distant organs.

The condition affects women primarily during their reproductive years and has become a major public health concern due to its high prevalence and substantial socioeconomic burden. Persistent pelvic pain, dysmenorrhea, dyspareunia, infertility, fatigue, and psychological distress significantly impair daily functioning and overall well-being. Many affected women experience years of symptoms before receiving an accurate diagnosis, resulting in delayed treatment and progression of disease-related complications.

Although endometriosis has been studied extensively for decades, its exact etiology remains controversial. Several theories have been proposed to explain disease development. The theory of retrograde menstruation suggests that menstrual debris flows backward through the fallopian tubes

into the pelvic cavity, where viable endometrial cells implant and proliferate. However, because retrograde menstruation occurs in many healthy women, additional factors are believed to contribute to disease establishment.

Genetic susceptibility has emerged as an important component of endometriosis pathogenesis. Family studies have demonstrated a significantly increased risk among first-degree relatives of affected individuals. Advances in genomic research have identified numerous genetic variants associated with inflammatory regulation, hormonal responsiveness, cellular adhesion, and tissue remodeling. Hormonal influences play a central role in disease progression. Endometriotic lesions exhibit enhanced sensitivity to estrogen and relative resistance to progesterone, creating a microenvironment that promotes cellular proliferation, angiogenesis, and chronic inflammation. These hormonal abnormalities contribute to lesion persistence and symptom development.

Immunological dysfunction has also been implicated in disease pathophysiology. Altered immune surveillance may permit ectopic endometrial cells to evade clearance mechanisms and survive within the peritoneal cavity. Increased production of inflammatory cytokines, growth factors, and oxidative stress mediators further supports lesion growth and chronic inflammatory responses.

Recent investigations have highlighted the importance of epigenetic regulation, stem cell activity, and environmental influences in disease development. Exposure to endocrine-disrupting chemicals and other environmental factors may interact with genetic predisposition to increase susceptibility in certain individuals. These findings support the concept that endometriosis results from a complex interaction between biological and environmental determinants.

Diagnosis of endometriosis remains challenging due to the heterogeneity of symptoms and the absence of a single definitive noninvasive diagnostic test. Clinical evaluation typically includes detailed symptom assessment, pelvic examination, ultrasonography, magnetic resonance imaging, and laboratory investigations. While laparoscopy has traditionally been considered the gold standard for diagnosis, advances in imaging technologies have improved noninvasive detection of deep infiltrating lesions and ovarian endometriomas.

Treatment strategies have evolved considerably in recent years. Contemporary management focuses on symptom relief, suppression of disease progression, preservation of fertility, and improvement of quality of life. Medical therapies include hormonal agents that reduce estrogen stimulation and control inflammatory activity. Surgical interventions are reserved for selected patients with severe symptoms, extensive disease, or infertility. Increasingly, multidisciplinary approaches incorporating pain management, reproductive medicine, nutrition, and psychological support are being recognized as essential components of comprehensive care.

Given the growing prevalence of endometriosis and its profound impact on women's health, continued investigation into disease mechanisms, diagnostic innovations, and therapeutic strategies remains critically important. The present study aims to evaluate modern perspectives on the pathogenesis, diagnosis, and treatment of endometriosis and to highlight current advances that may contribute to improved clinical outcomes.

2. Materials and Methods

This study was conducted between 2024 and 2025 in specialized gynecological and reproductive health centers. A total of 200 women aged 18–45 years who presented with symptoms suggestive of endometriosis were included in the investigation. Participants were selected based on clinical manifestations including chronic pelvic pain, dysmenorrhea, dyspareunia, infertility, abnormal uterine bleeding, and pelvic discomfort.

A comprehensive diagnostic protocol was applied to all participants. Detailed medical histories were obtained, including age at symptom onset, menstrual characteristics, reproductive history, previous gynecological interventions, family history of endometriosis, and associated chronic conditions. Clinical examination included abdominal and pelvic assessment performed by experienced gynecologists.

Diagnostic imaging consisted of transvaginal ultrasonography, transabdominal ultrasonography when indicated, and magnetic resonance imaging in patients suspected of having deep infiltrating endometriosis. Ovarian endometriomas, pelvic adhesions, and infiltrative lesions were documented and classified according to anatomical location and severity.

Laboratory investigations included complete blood count, inflammatory markers, hormonal profile assessment, and evaluation of serum biomarkers associated with endometriosis. In selected cases,

diagnostic laparoscopy was performed to confirm disease presence and determine lesion distribution. Histopathological examination was conducted whenever surgical tissue samples were obtained. Patients were categorized according to disease stage using internationally accepted classification systems. Treatment strategies were individualized based on symptom severity, disease extent, age, fertility intentions, and overall health status. Therapeutic interventions included hormonal therapy, anti-inflammatory medications, laparoscopic surgery, fertility-preserving procedures, and multidisciplinary supportive care. Clinical outcomes were evaluated over a twelve-month follow-up period. Primary endpoints included pain reduction, improvement in reproductive function, disease stabilization, quality of life enhancement, and patient satisfaction with treatment.

3. Results

The study demonstrated that chronic pelvic pain was the most frequently reported symptom among participants. Dysmenorrhea was present in a substantial proportion of women and was often associated with significant limitations in daily activities, educational performance, and occupational productivity. Many patients described progressive worsening of symptoms over several years before diagnosis was established.

Diagnostic imaging revealed ovarian endometriomas as one of the most common manifestations of the disease. Deep infiltrating lesions were identified in a considerable number of patients and frequently involved the uterosacral ligaments, rectovaginal septum, and pelvic peritoneum. Advanced-stage disease was often accompanied by extensive adhesions and distortion of normal pelvic anatomy.

A strong association was observed between disease severity and symptom burden. Women with extensive endometriotic lesions generally experienced more severe pelvic pain, greater functional limitations, and lower quality-of-life scores compared with those presenting with minimal or mild disease.

Hormonal treatment resulted in significant improvement in pain-related symptoms. Patients receiving hormonal suppression therapy demonstrated substantial reduction in dysmenorrhea, chronic pelvic pain, and dyspareunia during follow-up assessments. Symptom improvement was particularly evident among women who adhered consistently to prescribed treatment protocols.

Laparoscopic surgical management produced favorable outcomes in patients with moderate to severe disease. Excision of endometriotic lesions and restoration of normal pelvic anatomy contributed to meaningful reductions in pain intensity and improvement in reproductive function. Many women reported enhanced physical well-being and improved daily functioning following surgical intervention. Among participants experiencing infertility, treatment of endometriosis was associated with increased rates of spontaneous conception and improved outcomes following assisted reproductive procedures. Fertility improvement was most notable among women receiving early intervention before development of extensive pelvic damage.

Psychological assessment revealed that many patients experienced anxiety, emotional distress, and reduced social participation prior to treatment. Successful symptom management was accompanied by measurable improvements in mental well-being, interpersonal relationships, and overall quality of life.

4. Discussion

The findings of this study support the current understanding that endometriosis is a multifactorial disorder involving complex interactions among hormonal, immunological, inflammatory, genetic, and environmental mechanisms. The heterogeneous clinical presentation observed among participants reflects the biological complexity of the disease and contributes to challenges in diagnosis and management.

The high prevalence of chronic pelvic pain identified in this investigation confirms that pain remains one of the most significant clinical manifestations of endometriosis. Persistent inflammation, neuroangiogenesis, and sensitization of peripheral and central pain pathways likely contribute to the chronic nature of symptom development. These mechanisms explain why symptom severity does not always correlate directly with lesion size or disease stage.

The diagnostic delay frequently observed among affected women remains a major healthcare

concern. Many patients experience symptoms for years before receiving an accurate diagnosis. Increased awareness among healthcare professionals and broader utilization of modern imaging techniques may facilitate earlier recognition and reduce disease progression.

The effectiveness of hormonal therapy observed in this study highlights the central role of estrogen-dependent mechanisms in endometriosis pathophysiology. Suppression of hormonal stimulation reduces lesion activity and inflammatory responses, thereby alleviating symptoms and improving quality of life. However, recurrence following discontinuation of therapy remains a significant clinical challenge.

Surgical treatment continues to play an important role in carefully selected patients. Laparoscopic excision of lesions offers both diagnostic and therapeutic benefits and may significantly improve pain control and reproductive outcomes. Nevertheless, surgical management should be individualized due to the potential risks of recurrence and procedural complications.

An important finding of this investigation was the substantial psychosocial burden associated with endometriosis. Chronic pain, infertility, and prolonged diagnostic uncertainty often contribute to emotional distress and social impairment. These observations emphasize the need for multidisciplinary care models that address both physical and psychological aspects of the disease. Recent advances in molecular biology have expanded understanding of disease mechanisms and created opportunities for development of targeted therapies. Emerging treatments focusing on inflammatory pathways, angiogenesis regulation, immune modulation, and molecular signaling may provide additional therapeutic options in the future.

The study also reinforces the importance of fertility preservation in reproductive-age women. Early diagnosis and timely intervention may reduce reproductive complications and improve long-term fertility outcomes. Collaboration between gynecologists, reproductive specialists, pain management experts, and mental health professionals is essential for comprehensive patient care.

5. Conclusion

Endometriosis is a chronic and multifaceted gynecological disorder that significantly affects reproductive health, physical functioning, psychological well-being, and overall quality of life. The disease arises through complex interactions among hormonal, inflammatory, immunological, genetic, and environmental factors, making diagnosis and treatment particularly challenging.

Modern diagnostic approaches, including advanced imaging technologies and minimally invasive surgical techniques, have improved the accuracy of disease detection and staging. Early recognition remains crucial for preventing disease progression and reducing long-term complications.

The findings of this study demonstrate that individualized treatment strategies combining hormonal therapy, surgical intervention when necessary, fertility management, and psychosocial support can significantly improve clinical outcomes. Effective management not only reduces symptom severity but also enhances reproductive potential and overall patient well-being.

Future research should focus on identifying reliable biomarkers, improving noninvasive diagnostic methods, and developing targeted therapies that address underlying disease mechanisms. Continued advances in personalized medicine are expected to further optimize the care of women affected by endometriosis and improve long-term health outcomes.

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