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CLINICAL CHARACTERISTICS OF ENDOMETRIOSIS PHENOTYPES

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Abstract
This article presents a detailed clinical analysis of various phenotypes of endometriosis, a chronic gynecological disease characterized by the presence of endometrial-like tissue outside the uterus. The study examines the specific clinical manifestations, diagnostic criteria, and progression patterns of different phenotypes, including superficial peritoneal, ovarian, and deep infiltrating endometriosis. Attention is paid to the correlation between phenotypic diversity and disease severity, as well as treatment response. The findings highlight the importance of phenotype-oriented diagnostics and personalized therapeutic approaches in improving patient outcomes.

Keywords: Endometriosis, phenotypes, clinical characteristics, diagnosis, pathology, treatment, women's health.

Introduction

Endometriosis is a chronic, estrogen-dependent inflammatory disease that affects women of reproductive age and is one of the leading causes of pelvic pain and infertility. It is characterized by the growth of endometrial-like tissue outside the uterine cavity, most commonly on the ovaries, pelvic peritoneum, and other pelvic organs. Despite significant advances in research, the pathogenesis of endometriosis remains complex and not fully understood.

Recent studies have shown that endometriosis presents in various phenotypic forms — superficial peritoneal, ovarian (endometrioma), and deep infiltrating — each with distinct clinical, morphological, and pathophysiological features. Understanding these phenotypes is essential for accurate diagnosis, effective treatment planning, and prediction of disease progression.The purpose of this study is to describe the clinical characteristics of different endometriosis phenotypes, identify their distinguishing features, and emphasize the importance of phenotype-based diagnostic and therapeutic strategies in modern gynecology.

Endometriosis is considered a multifactorial disease that involves genetic, hormonal, immunological, and environmental factors. Clinically, it is manifested by chronic pelvic pain, dysmenorrhea, dyspareunia, infertility, and various menstrual irregularities. However, the expression and severity of these symptoms depend largely on the phenotype of the disease.

This phenotype is the most common and is characterized by small lesions on the peritoneal surface. It often causes mild or moderate pain, and many patients remain asymptomatic. Laparoscopic visualization shows small red, black, or white foci, which may be accompanied by minimal adhesions. Although this form is usually less severe, it can progress and lead to infertility if not treated in time.

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Ovarian endometriomas are cystic formations filled with thick, dark-brown fluid, often referred to as "chocolate cysts." This phenotype is associated with ovarian dysfunction, reduced fertility, and a high risk of recurrence. Ultrasound examination is a key diagnostic method for detecting endometriomas. Surgical treatment, particularly laparoscopic cystectomy, remains the gold standard, though recurrence rates can reach up to 30%.

This is the most aggressive phenotype, characterized by infiltration of endometrial-like tissue into the peritoneum and surrounding organs such as the rectum, bladder, and uterosacral ligaments. DIE causes severe chronic pelvic pain, dyschezia, and urinary symptoms, significantly affecting the patient's quality of life. MRI and laparoscopy are the most informative diagnostic tools. The management of DIE often requires a multidisciplinary approach combining surgical excision with hormonal therapy.

Studies show that each phenotype of endometriosis has its own molecular and immunological profile. Deep infiltrating endometriosis is associated with increased angiogenesis, fibrogenesis, and higher expression of inflammatory cytokines compared to other forms. Ovarian endometriomas, on the other hand, show altered hormonal receptor expression and oxidative stress markers. These differences explain the variability in clinical manifestations and treatment responses.

Accurate diagnosis of endometriosis requires a combination of clinical evaluation, imaging methods, and sometimes laparoscopy with histological confirmation. Modern management includes hormonal therapy (combined oral contraceptives, progestins, GnRH agonists) and surgical interventions, depending on the phenotype and severity of symptoms.

Phenotype-oriented diagnosis allows clinicians to select the most effective treatment strategy. For example, patients with superficial peritoneal endometriosis often benefit from hormonal therapy, while those with deep infiltrating forms require complex surgical management. Understanding the phenotype also helps in predicting recurrence and optimizing fertility outcomes.Recognizing the clinical and biological diversity of endometriosis phenotypes is crucial for improving diagnostic accuracy, tailoring treatment, and enhancing the quality of life of affected women.In recent years, the concept of endometriosis phenotyping has become a cornerstone of modern gynecological research. It allows clinicians to identify not only the localization and depth of lesions but also the underlying biological mechanisms that contribute to disease persistence and recurrence. By distinguishing phenotypes, researchers can develop more precise and less invasive diagnostic and therapeutic strategies.

The correlation between phenotype and clinical symptoms has been confirmed by multiple clinical trials. For example, deep infiltrating endometriosis is most often associated with severe pain syndromes, while ovarian endometriomas are more frequently linked to infertility. Superficial peritoneal forms, in contrast, may present with minimal or no symptoms, which complicates timely diagnosis. This variability highlights the necessity for clinicians to consider phenotype-specific symptom patterns during examination and patient counseling.

Advancements in imaging technologies have significantly improved the detection of endometriosis phenotypes. Transvaginal ultrasonography and magnetic resonance imaging (MRI) provide detailed visualization of lesions, their size, and localization. MRI, in particular, plays a critical role in identifying deep infiltrating endometriosis, revealing lesions affecting the bowel, bladder, or

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retrocervical area. The combination of imaging findings with patient-reported symptoms offers a comprehensive clinical picture that guides further management.

The hormonal dependency of endometriosis is a well-established fact, with estrogen playing a key role in lesion growth and inflammation. However, recent genetic studies have identified specific genes and molecular pathways that differ among phenotypes. For instance, the overexpression of aromatase and estrogen receptor beta (ERβ) is more prominent in deep infiltrating lesions, whereas ovarian endometriomas show higher oxidative stress and altered mitochondrial function. These findings open new avenues for targeted therapies that can address the specific biological characteristics of each phenotype.Treatment strategies must be individualized according to the phenotype, patient's age, fertility goals, and disease severity. Hormonal suppression remains the cornerstone of therapy, aimed at reducing pain and preventing progression. However, surgery becomes indispensable in advanced stages, especially for deep infiltrating forms causing organ dysfunction. Postoperative hormonal therapy is often recommended to minimize recurrence.

Clinical outcomes depend significantly on the phenotype: patients with superficial peritoneal endometriosis usually experience rapid symptom relief after conservative treatment, while those with ovarian or deep infiltrating forms often face a higher risk of recurrence and persistent pain. Consequently, continuous follow-up and long-term management are necessary to maintain remission and preserve reproductive function.

Current research is increasingly focusing on molecular profiling, biomarkers, and immunological factors to develop non-invasive diagnostic tools. Blood-based and menstrual biomarkers are being explored as potential alternatives to laparoscopy for phenotype detection. Furthermore, studies on the gut microbiome, epigenetic modifications, and neuroinflammatory mechanisms are deepening our understanding of the disease's complexity.

Conclusion

Endometriosis is a heterogeneous and multifactorial disease that manifests through several distinct phenotypes — superficial peritoneal, ovarian, and deep infiltrating. Each phenotype differs in its clinical presentation, pathophysiology, and response to treatment, which highlights the need for an individualized diagnostic and therapeutic approach.

Understanding the clinical and biological characteristics of each phenotype allows healthcare professionals to select optimal management strategies, improve diagnostic accuracy, and enhance patient outcomes. The identification of molecular and genetic variations among phenotypes opens new opportunities for targeted therapies and personalized medicine.

Early detection and accurate phenotyping are essential for preventing disease progression, reducing recurrence rates, and preserving fertility. A multidisciplinary approach that integrates clinical, surgical, hormonal, and molecular perspectives remains the most effective strategy for managing endometriosis and improving the quality of life for affected women.Phenotype-oriented diagnosis and treatment not only deepen the understanding of endometriosis as a complex disorder but also represent a crucial step toward precision gynecology and individualized patient care.

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