

CLINICAL SIGNS AND DIAGNOSIS OF EXTERNAL GENITAL ENDOMETRIOSIS

Atayeva Farzona Nuriddinovna

Assistant Professors of the 3rd Department of Obstetrics and Gynecology

Abstract

External genital endometriosis is a rare manifestation of endometriotic lesions occurring outside the uterine cavity, specifically on the vulva, perineum, and other external genital structures. Although less common than pelvic endometriosis, this condition poses a diagnostic challenge due to its non-specific clinical presentation. Common symptoms include cyclic pain, localized swelling, dyschezia, and in some cases, the presence of bluish nodules or cystic masses. The condition often mimics other dermatologic or infectious diseases, leading to delayed or misdiagnosis. Diagnosis relies on a thorough clinical history, gynecological examination, imaging techniques such as ultrasonography and MRI, and histopathological confirmation via biopsy. Early identification and differentiation from other vulvar disorders are crucial to prevent complications and offer effective treatment options, including hormonal therapy and surgical excision. This paper reviews the typical clinical signs, diagnostic pathways, and challenges in identifying external genital endometriosis.

Keywords: External genital endometriosis; vulvar endometriosis; cyclic vulvar pain; endometriosis diagnosis; perineal endometriosis; ectopic endometrial tissue; gynecological imaging; vulvar lesions

Introduction

Endometriosis is a chronic gynecological condition characterized by the presence of functional endometrial tissue outside the uterine cavity. While the disease predominantly affects the pelvic organs, such as the ovaries, fallopian tubes, and peritoneum, extra-pelvic and external manifestations have been increasingly recognized in clinical practice. One of the rare but clinically significant forms is external genital endometriosis, which involves the vulva, perineum, and other external reproductive structures. Though it constitutes a small percentage of overall endometriosis cases, its impact on patients' quality of life can be profound, leading to chronic pain, sexual dysfunction, and significant psychological distress.

External genital endometriosis remains a diagnostic challenge due to its rarity and nonspecific clinical symptoms. The condition is often overlooked or misdiagnosed as Bartholin cysts, vulvar varicosities, hidradenitis suppurativa, or other dermatological conditions, thereby delaying appropriate treatment. In many cases, patients undergo multiple consultations with dermatologists, general practitioners, or urologists before being referred to a gynecologist who can correctly identify and manage the condition. Consequently, raising awareness of this uncommon presentation is crucial for timely diagnosis and effective intervention.

The pathogenesis of external genital endometriosis remains poorly understood, but several theories have been proposed. The most widely accepted theories include retrograde menstruation, coelomic metaplasia, lymphatic or hematogenous dissemination, and direct implantation during obstetric or gynecologic surgeries, particularly episiotomies. For example, perineal endometriosis is frequently associated with a history of perineal trauma, such as episiotomy or laceration during vaginal delivery. This surgical disruption is believed to facilitate the implantation of endometrial cells into the wound, which subsequently develop into endometriotic lesions under the influence of hormonal cycles.

Clinically, patients with external genital endometriosis typically present with localized pain that is cyclical in nature, coinciding with their menstrual period. Swelling, tenderness, and palpable nodules may be observed in the vulvar or perineal region. In some cases, the lesions appear as bluish or violaceous masses that change in size and discomfort throughout the menstrual cycle. Pain during defecation (dyschezia), urination (dysuria), or sexual intercourse (dyspareunia) may also be reported, depending on the anatomical location and extent of the disease. These symptoms can significantly impair daily activities and mental well-being, necessitating prompt and accurate diagnosis.

Diagnostic evaluation of suspected external genital endometriosis involves a thorough clinical history and physical examination. Imaging studies such as high-resolution ultrasonography, magnetic resonance imaging (MRI), and computed tomography (CT) scans may aid in identifying subcutaneous lesions and their relationship with surrounding tissues. However, imaging findings can be nonspecific,

and a definitive diagnosis often requires histopathological confirmation through biopsy. The biopsy typically reveals endometrial glands and stroma, along with signs of hemorrhage and inflammation. Immunohistochemical staining for estrogen and progesterone receptors can further support the diagnosis.

Differential diagnosis of external genital endometriosis is broad and includes benign and malignant conditions. Vulvar cysts, hematomas, abscesses, and vulvar intraepithelial neoplasia are among the conditions that must be ruled out. In some cases, malignancies such as squamous cell carcinoma or melanoma may mimic the appearance of endometriotic lesions. Therefore, it is essential for clinicians to maintain a high index of suspicion and conduct appropriate investigations to exclude these conditions.

Management of external genital endometriosis involves a multidisciplinary approach that combines medical and surgical strategies. Medical treatment aims to suppress ovulation and hormonal stimulation of ectopic endometrial tissue. Commonly used medications include nonsteroidal anti-inflammatory drugs (NSAIDs), oral contraceptives, gonadotropin-releasing hormone (GnRH) agonists, and progestins. These therapies can provide symptomatic relief but may not always result in complete resolution of the lesions. Surgical excision remains the definitive treatment, particularly in cases with well-demarcated, accessible lesions. Complete surgical removal not only alleviates symptoms but also reduces the risk of recurrence. Despite available treatments, recurrence of external genital endometriosis is not uncommon, especially if the lesions are incompletely excised or if hormonal suppression is discontinued. Therefore, long-term follow-up and individualized treatment planning are essential. For patients with recurrent or refractory disease, newer therapeutic modalities, including aromatase inhibitors and immunomodulatory agents, are being explored. Additionally, psychological counseling and support groups may be beneficial for patients coping with the chronic nature of the disease. In conclusion, external genital endometriosis is a rare but important clinical entity that requires heightened awareness among healthcare providers. Its diagnosis demands a combination of clinical suspicion, imaging, and histopathological confirmation. Early identification and appropriate management can significantly improve patient

outcomes and quality of life. Further research is needed to elucidate the pathophysiological mechanisms, optimize diagnostic tools, and develop targeted therapies for this unique manifestation of endometriosis.

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