Diagnosing endometriosis in primary care: clinical update

INTRODUCTION
Prompt diagnosis of endometriosis is a clinical imperative for general practice. Women with endometriosis typically face a diagnostic delay of 7–10 years, despite more frequent GP and accident and emergency attendances than women without the disease.1,2 Endometriosis has a relatively high prevalence (up to 10% of women in the general UK population and up to 50% of infertile women) and causes high morbidity in terms of both pain and infertility.2 Despite this, the condition remains poorly recognised in practice, meaning that patients must often navigate misdiagnosis and suboptimal care. While there have been recent significant advances in the evidence regarding pathogenesis and management, this literature is largely directed towards a secondary care audience. We present key issues below from a primary care perspective.

REASONS FOR DIAGNOSTIC DELAY
Diversity of presenting symptoms, overlap with benign conditions, and a low index of suspicion in both primary and secondary care all contribute to women with endometriosis falling between the cracks. Other contributors include cultural attitudes normalising painful menstruation, a lack of clinician awareness of updated guidelines on distinguishing normal from pathological, and concern over the invasive nature of laparoscopy.1,2 Paradoxically, while women presenting with multiple symptoms are more likely to have endometriosis,3 frequent attendance with disparate symptoms may make diagnosis less likely, as patients are dismissed as having a functional or psychosomatic cause.1

RECOGNISING ENDOMETRIOSIS
Endometriosis is a chronic inflammatory condition, in which deposits of ectopic endometrial-type tissue cause cyclical bleeding, scarring, and adhesion formation throughout the pelvis and sometimes beyond. Symptoms begin as early as adolescence and typically settle after menopause. There appears to be a genetic link, with many women reporting a first-degree relative with the disease.1

Cardinal symptoms are dysmenorrhoea, dyspareunia, and chronic (non-menstrual) pelvic pain. Bowel symptoms (painful defaecation or irritable bowel-type dysfunction) are common, even in the absence of overt bowel involvement. Back pain, fatigue, bladder symptoms (two-thirds have associated interstitial cystitis) and abnormal bleeding (heavy menstrual bleeding, pre-menstrual spotting, or postcoital bleeding) are also commonly reported. Pelvic pain that comes on before the onset of bleeding raises the level of clinical concern. Presentation may be esoteric: any symptom which reliably worsens with the menstrual cycle should prompt consideration of the diagnosis.2,3

Associated conditions include atopic and autoimmune disorders, inflammatory bowel disease, and interstitial cystitis. There is also an increased risk of ovarian carcinoma.3

PRACTICE POINTS: HISTORY, EXAMINATION, AND INVESTIGATION
Women with endometriosis attend the GP more frequently, offering crucial opportunities for diagnosis.5 In women presenting in primary care, a careful history is essential and will often suggest the diagnosis. Importantly, morbidity does not necessarily correlate with severity; women with minimal disease at laparoscopy may be highly symptomatic, and vice versa.

• Ask specifically about menarche, history of menstrual difficulties, details of pregnancies, and any difficulties with conception.
• Ask also about dysmenorrhoea, dyspareunia, pelvic pain throughout the cycle, and cyclical bowel and bladder symptoms.

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• Consider asking women presenting primarily with irritable bowel syndrome (IBS) symptoms about their menstrual history.

Examination findings such as pain, vaginal nodules, or adnexal masses support a clinical suspicion of endometriosis, but a normal examination does not rule out endometriosis.2

Primary care investigations such as ultrasound or CA-125 are not reliable indicators of the presence or absence of disease. Patients with suspected endometriosis should be referred to a specialist for consideration of laparoscopy. Women should ideally be referred to a tertiary endometriosis centre where available, or to a surgeon with a special interest where it is not.2,3

DIAGNOSTIC PITFALLS
Prompt diagnosis ensures appropriate care. Clinical suspicion should prompt referral. Empirical management with the contraceptive pill is common practice in primary care. Periods can often effectively be eliminated by continuous use of either a combined pill [running packets back to back], or a progesterone-only pill that reliably suppresses ovulation, such as desogestrel (Cerazette®, MSD). This offers a useful and practical way to control symptoms, but should not prevent or delay referral for definitive diagnosis.2 On the other hand, empirical treatment that may offer symptomatic relief does not need to be withheld while awaiting surgical confirmation of disease.4

Women with severe endometriosis should be referred to a specialist multidisciplinary centre. This ensures access to gynaecologists, bowel surgeons, and urologists skilled in laparoscopic management, as well as specialist nurses, psychology services, and an active network of support groups.4

Contrary to traditional teaching, endometriosis does occur in adolescents and younger women. Indeed, it has been reported that most women with endometriosis first experienced symptoms in their teens.1 Although this group are often highly symptomatic, diagnosis may be delayed by a cultural belief among both doctors and patients that painful periods in this age group are normal.3

• Ask about the impact of pain on normal activity, and have a high index of suspicion where pain is debilitating or regularly associated with vomiting.

• Age should not be a factor in deciding whether to refer.

Bowel and bladder symptoms are easily missed. Women with endometriosis are more likely to be diagnosed with IBS, and this may represent a missed diagnosis rather than comorbidity. Other common pitfalls are a misdiagnosis of ovarian cyst or pelvic inflammatory disease. Women presenting with multiple symptoms over time are at higher risk of endometriosis.2 Have a high index of suspicion and a holistic approach in approaching such patients.

CONCLUSION
Long diagnostic delays, misdiagnoses, and subsequent inappropriate management mean that women with endometriosis are often being failed by healthcare services. GPs are in a strong position to advocate for this underserved group of patients. Awareness of common presentations, a high index of clinical suspicion, and early referral should help improve awareness of this common and debilitating illness.

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