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Diagnosing diagnostic error of endometriosis: a secondary analysis of patient experiences from a mixed-methods survey

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ABSTRACT

Objective To analyse endometriosis diagnostic errors made by clinicians as reported by patients with endometriosis.

Methods This study deductively analysed qualitative data as part of a larger mixed-methods research study examining 'invalidating communication' by clinicians concerning patients' symptoms. Data analysed were responses to an open-ended prompt asking participants to describe an interaction with a clinician prior to their diagnosis in which they felt their symptoms were dismissed. We used three validated taxonomies for diagnosing diagnostic error (Diagnosis Error Evaluation and Research (DEER), Reliable Diagnosis Challenges (RDC) and generic diagnostic pitfalls taxonomies).

Results A total of 476 relevant interactions with clinicians were identified from 444 patients to the open-ended prompt, which identified 692 codable units using the DEER taxonomy, 286 codable units using the RDC taxonomy and 602 codable diagnostic pitfalls. Most prevalent subcategories among these three taxonomies were inaccurate/misinterpreted/overlooked critical piece of history data (from DEER Taxonomy; n=291), no specific diagnosis was ever made (from diagnostic pitfalls taxonomy; n=271), and unfamiliar with endometriosis (from RDC Taxonomy; n=144).

Conclusion Examining a series of patient-described diagnostic errors reported by patients with surgically confirmed endometriosis using three validated taxonomies demonstrates numerous areas for improvement. These findings can help patients, clinicians and healthcare organisations better anticipate errors in endometriosis diagnosis and design and implement education efforts and safety to prevent or mitigate such errors.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Diagnostic delay of endometriosis is well documented, and studies have been limited to qualitative studies with small sample sizes.

WHAT THIS STUDY ADDS

⇒ The key shortcomings in achieving timely and optimal diagnosis were seen in both general practitioners as well as obstetric and gynaecology generalists, both of whom often erred in dismissing patients symptoms as 'normal' menstrual pain and failed to order recommended diagnostic procedures.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ These findings can help patients, clinicians and healthcare organisations design and implement education efforts and safety nets for the anticipation, prevention and/or mitigation of such errors

INTRODUCTION

Recent patient safety efforts have focused on diagnostic error, defined as 'the failure to (a) establish an accurate and timely explanation of the patient's health problem(s) or (b) communicate that explanation to the patient'.[1 2] Diagnostic errors have an incidence of 10–15% in the general medical field and are the most common and costly reason for medical malpractice claims and contribute significantly to preventable patient harm.[3–6]

One diagnosis that deserves special attention in people assigned female at birth is endometriosis, the most common non-malignant gynaecological disease. Endometriosis is broadly defined as the presence of endometrial-like tissue in ectopic location.[7] Common symptoms include chronic pelvic pain, dysmenorrhoea, dyspareunia, fatigue and infertility.[7 8] Endometriosis is diagnosed in roughly 10% of people assigned female at birth—roughly equivalent to the prevalence of type 2 diabetes in the USA.[9 10] Endometriosis is associated with significant physical and psychological morbidity and contributes to economic burden.[11 12] Currently, endometriosis can be definitively diagnosed only through surgical visualisation and histological confirmation of biopsied lesions, though there is emerging evidence that advanced ultrasonography can be used as a diagnostic tool by trained specialists in endometriosis.[13 14]

Despite its high prevalence, diagnostic error is pervasive in endometriosis, being described

as 'a common and commonly missed and delayed diagnosis'.[15] 75.2% of non-randomly sampled US patients with endometriosis reported having had their endometriosis misdiagnosed, with a mean diagnostic delay ranging from 8 to 12 years.[16 17] A recent systematic review of qualitative studies identified reasons for diagnostic delay and included patient factors, social factors, clinician factors and disease-specific factors.[18] Patient factors include difficulty differentiating pathological symptoms from normal menstruation and self-care techniques.[18] Social factors include menstrual stigma and normalisation of menstrual pain by other females.[18] Clinician and health system factors include dismissal of endometriosis symptoms by clinicians (ie, symptom invalidation[19]), clinicians' poor knowledge of endometriosis, delayed referral to specialist services and poor explanation of the contraceptive pill in the diagnostic process (ie, clinicians using response to oral contraceptives as a diagnostic criterion).[19–21] Disease-specific factors include variability in presenting symptoms and overlap with other conditions, lack of a non-invasive method of definitive diagnosis and concerns about the value of reaching a definitive diagnosis.[18]

Despite the prevalence of diagnostic error in endometriosis, there has been limited research to elucidate reasons contributing to these errors. Of the research that does exist,[21] studies have been limited to qualitative studies with, correspondingly, small sample sizes. The present study thus takes a systematic approach to codifying reasons for diagnostic error in endometriosis, using a large sample of patients with endometriosis, by deductively analysing qualitative data as part of a large mixed-methods research study using taxonomies developed specifically for analysing diagnostic error under the guidance of a diagnostic safety expert.

METHODS

This study deductively analysed and subsequently quantified qualitative data as part of a larger online mixed-methods research study that sought to understand patients' experiences with symptom invalidation from a clinician and develop a patient-reported measure of symptom invalidation experienced within a single interaction.

Participants

Inclusion criteria were (1) self-reported diagnosis of endometriosis, (2) self-reported age of 18+ years, and (3) English proficiency. For this paper, we excluded patients without a outside the USA.

Data collection

Data were collected between March and June 2019. Patients were recruited from an advertisement posted to social media sites (eg, Facebook, Instagram) hosted by endometriosis non-profit organisations (eg, Endometriosis Research Center, EndoWhat?, Endometriosis Foundation of America). The advertisement described the study as research to learn about potential 'dismissal or invalidation of your medical symptoms that you may have experienced by healthcare providers and/or your romantic partner, and how this invalidation may be related to your physical and mental health'. This larger study thus purposively recruited patients who have perceived having experienced symptom invalidation. Those who accessed the survey link were brought to a page that included the digital consent form, which after agreeing to participate took them to a 20–30 min survey.

Following entering demographic, health-related information and items inquiring about their healthcare interactions, participants were provided with the following open-ended prompt:

> Please describe a specific interaction you had with a doctor that took place *before your diagnosis of endometriosis* in which your doctor said, implied, and/or did one or more things that you felt were dismissive of your endometriosis symptoms and/or you. As best you can, please include specifically what the doctor said. If there has been more than one interaction, please choose the most memorable.

After describing a specific healthcare interaction, for the purposes of the larger study that sought to understand patients' experiences with symptom invalidation from a clinician and develop a patient-reported measure of symptom invalidation experienced within a single interaction, patients completed a series of close-ended questions pertaining to the specific interaction they described. The self-report scale of symptom invalidation asked them to reflect on the specific interaction, and the study included questionnaires on patient satisfaction, supportive message quality and cognitive/affective reactions to the clinician-patient interaction. Social support, self-esteem and depression were also assessed.

Establishing sample size and units of analysis

1747 patients consented to the survey and 1631 initiated the survey. 1041 (63.9%) patients from the USA and abroad inserted some text into the text field following the open-ended prompt, though 26 (2.5%) did not respond appropriately to the prompt (eg, typed '1', 'None', 'Nothing before diagnosis', 'I have had this happen to me'). Among those who appropriately responded to the prompt (n=1041), 123 (11.8%) were excluded because they did not report having a surgically confirmed diagnosis of endometriosis, yielding 918 patients. Another 333 (36.3%) from the remaining 918 were excluded because they did not report being from the USA. The responses of the remaining 585 patients were then analysed to determine if their response provided details of a diagnostic error as defined by the National Academy of Medicine, which yielded 444 relevant responses.[2] Of the 141 discarded responses of the 585 patients, 11 (7.8%) reported that they did not experience symptom invalidation.

Not all patients adhered to the instructions to describe only one interaction. A total of 365 patients (82.6%)

described one (or more) interaction with one clinician; however, 22 patients (5.0%) described interactions with two clinicians, 2 (0.5%) described interactions with three, 1 (0.2%) described interactions with four, 1 (0.2%) described interactions with six, and 51 (11.6%) described general interactions with clinicians (eg, 'Most doctors said…'). Two patients (0.5%) did not name a specific type of clinician, but instead described they did not know their symptoms were abnormal since they began at menarche, so they did not seek out a clinician (which contributes to diagnostic delay).[21] Because 79 patients described an interaction with more than one clinician, the unit of analysis was not the number of patient responses but instead the number of clinicians with whom patients described interactions. For the 51 patients who described interactions with clinicians generally, their responses were coded as a single unit of analysis, and clinician type was recoded as 'unspecified' because there was no way to distinguish the number of interactions or clinician type. Thus, there were thus 476 interactions reported (ie, 476 units of analysis). Within one unit of analysis, more than one reported factor contributing to diagnostic error could be coded.

Data analysis

Diagnostic errors were deductively analysed using three complementary taxonomies for diagnosing diagnostic errors and subsequently quantified. One, the Diagnosis Error Evaluation and Research (DEER) Taxonomy, identifies *where* a failure may have occurred in the diagnostic process.[22 23] The second, the Reliable Diagnosis Challenges (RDC) Taxonomy, identifies general challenges in the diagnostic process that may have contributed to the error/delay (ie, *why* an error may have occurred).[24] The third, a taxonomy for diagnostic pitfalls, classifies generic clinical situations that are susceptible to errors that may lead to missed, delayed or wrong diagnoses.[25] Although there are overlaps in various domains covered in each of the three taxonomies, they independently coded differing constructs. The three taxonomies are used separately, as they complement each other (see online supplemental eTable 6).

The first author, a postdoctoral research fellow with a PhD in health communication who is also a patient with endometriosis collected these data during her PhD programme. She reviewed all the data independently. She then met with the second author, a diagnosis expert, to discuss selected cases where clinical or classification questions arose and to discuss implications of the results. We used NVivo V.14 to qualitatively code the data and SPSS to quantify these responses and categorise them by clinician type.[26 27] Descriptive statistics were calculated based on categories organised by clinician type and were illustrated using histograms. Demographic and health-related characteristics were analysed descriptively using SPSS.[27]

Patient involvement

The first author, a patient with endometriosis, led the design and conduct of this research. Endometriosis organisations, many headed by individuals with endometriosis themselves, recruited subjects via their organisation's social media site(s). Once this study has been published, participants will be informed of the results via email and via dissemination by endometriosis-related organisations.

RESULTS

Patient demographic and health-related characteristics

Table 1 presents demographic and health-related characteristics. Of the 444 patients, mean reported age was 34.4, with 85.6% identifying as non-Hispanic white. Most reported being heterosexual (85.4%). The sample was generally well educated—64.5% reported having at least an undergraduate degree. Further, 76.7% reported an income of greater than $40 000.

Symptoms endorsed by patients are presented in online supplemental eFigure 1. Average time since diagnosis was 6.1 years. Average time from symptom onset to presentation to a clinician was 4.1 years. The average time from diagnosis seeking to surgical diagnosis was 6.5 years. 223 (50.2%) patients indicated they received clinical diagnosis prior to surgical diagnosis. Average time from diagnosis seeking to clinical diagnosis was 5.0 years, and average time from clinical diagnosis to surgical diagnosis was 1.7 years. Average diagnostic delay (ie, symptom onset to surgical diagnosis) was 11.1 years.

Diagnostic error clinician type

Of the 476 interactions reported, 16.6% were with general practitioners (GPs), 55.9% were with obstetricians/gynaecologists (OB/GYNs), 3.8% were with nurse practitioners (NPs), 5.9% were with emergency department physicians, 6.1% were with other types of clinicians and 13.4% were unspecified.

Diagnostic error analysis results

DEER Taxonomy categories

Of the 476 reported interactions with clinicians, 692 specific DEER Taxonomy items were coded. Based on the seven main DEER Taxonomy categories, 3 (0.4%) errors were associated with *access/presentation*, 344 (49.7%) were associated with the *history*, 7 (1.0%) were associated with the *physical exam*, 143 (20.7%) were associated with *tests*, 169 (24.4%) were associated with *assessment*, 23 (3.3%) were associated with *referral/consultation* and 3 (0.4%) errors were associated with *follow-up*. A breakdown of these DEER Taxonomy categories per clinician type is given in online supplemental eTable 1 and online supplemental eFigure 2.

The five most prevalent DEER Taxonomy subcategories were inaccurate/misinterpreted/overlooked critical piece of history data (*history* main category; n=291, 42.9%), too much weight on competing/coexisting diagnosis (*assessment* main category; n=87, 12.6%), failure/delay in ordering needed tests (*tests* main category; n=70, 10.1%), failure/delay in considering the diagnosis (*assessment* main category; n=53, 7.7%) and

Table 1 Sample demographic and health-related characteristics of respondents with included responses (n=444)

Characteristics	N (%)
Demographic characteristics	
Age (years)	M=34.4 (SD=8.0)
Race/ethnicity	
Hispanic Asian	1 (0.1%)
Hispanic Native Hawaiian or Pacific Islander	1 (0.1%)
Hispanic white	9 (2.0%)
Non-Hispanic American Indian/ Alaskan Native	2 (0.5%)
Non-Hispanic Asian	9 (2.0%)
Non-Hispanic black or African American	5 (1.1%)
Non-Hispanic white	380 (85.6%)
Non-Hispanic mixed race	15 (3.4%)
Missing	22 (5.0%)
Sexual orientation	
Heterosexual	379 (85.4%)
Lesbian	9 (2.0%)
Bisexual	37 (8.3%)
Queer	7 (1.6%)
Other	4 (1.8%)
Missing	8 (1.8%)
Education	
Some high school	1 (0.2%)
High school diploma or equivalent	31 (7.0%)
Business/trade/technical school	11 (2.5%)
Some college/2-year degree	107 (24.1%)
College degree	161 (36.3%)
Graduate degree	125 (28.2%)
Missing	8 (1.8%)
Yearly income >$40 000	
<$10 000	23 (5.2%)
$10 000 to $25 000	28 (6.3%)
$25 001 to $40 000	49 (11.0%)
$40 001 to $75 000	121 (27.3%)
$75 001 to $100 000	80 (18.0%)
$100 001 to $150 000	83 (18.7%)
>$150 000	46 (10.4%)
Missing	14 (3.2%)
Time since diagnosis (years)	M=6.1 (SD=6.5)
Time from symptom onset to diagnosis seeking (years)	M=4.1 (SD=5.3)
Time from diagnosis seeking to surgical diagnosis (years)	M=6.5 (SD=6.3)
Clinical diagnosis prior to surgical diagnosis (yes)	223 (50.2%)
Time from diagnosis seeking to a clinical diagnosis (years)	M=5.0 (SD=5.0)
Time from clinical diagnosis to surgical diagnosis (years)	M=1.7 (SD=3.8)
Diagnostic delay (years)	M=11.1 (SD=6.9)
Endometriosis-associated infertility	
Yes	174 (39.2%)
No	60 (13.5%)
Don't know	209 (47.1%)
Missing	1 (0.2%)
Average level of pain past 7 days (0–100)	M=46.8 (SD=24.7)
Average level of fatigue past 7 days (0–100)	M=63.5 (SD=23.5)
Average level of worst symptom past 7 days (0–100)	M=56.9 (SD=28.2)
Number of days health interference (0–30)	M=10.5 (SD=8.7)
Self-rated health (1–5)	M=2.7 (SD=0.9)

error in clinician interpretation of test or report (*tests* main category; n=36, 5.2%). A breakdown of these most prevalent DEER Taxonomy categories per clinician type is given in online supplemental eTable 2 and figure 1 and patient exemplars are in online supplemental eTable 7.

RDC Taxonomy categories

Of the 476 interactions, 286 specific RDC Taxonomy categories were identified. Based on the five main RDC Taxonomy categories, 159 (55.6%) errors were associated with a *challenging disease presentation*, 23 (8.0%) were associated with the *patient factors*, 66 (23.1%) were associated with the *testing challenges*, 1 (0.3%) was associated with *stressors* and 37 (12.9%) were associated with *broader challenges/difficulties*. A breakdown of the main RDC Taxonomy categories per clinician type is given in online supplemental eTable 3 and online supplemental eFigure 3.

The five most prevalent RDC Taxonomy subcategories were unfamiliar (*challenging disease presentation* main category; n=114, 39.9%); knowledge, complexities, test selection, ordering challenges (*testing challenges* main category; n=26, 9.1%); false negative results (*testing challenges* main category; n=23, 8.0%); recognising failure to respond to treatment (*broader challenges/difficulties* main category; n=20, 7.0%) and patient failure to follow-up (*patient factors* main category; n=16, 5.6%). A breakdown of the most prevalent RDC Taxonomy subcategories per clinician type is given in online supplemental eTable 4 and figure 2 and patient exemplars are in online supplemental eTable 7.

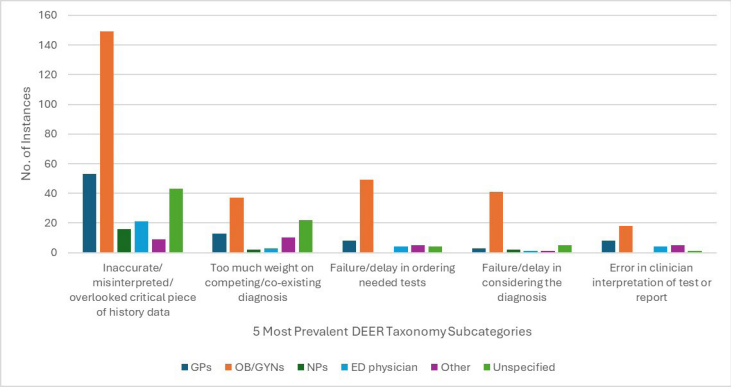

Figure 1 Five most prevalent DEER Taxonomy subcategories by clinician type. DEER, Diagnosis Error Evaluation and Research; ED, emergency department; GPs, general practitioners; NPs, nurse practitioners; OB/GYNs, obstetricians/gynaecologists.

Diagnostic pitfall taxonomy categories

Of the 476 interactions, 602 specific diagnostic pitfalls taxonomy categories were identified. Based on the six main diagnostic pitfalls taxonomy categories, 455 (75.6%) pitfalls were associated with *diagnosis/assessment*, 20 (3.3%) were associated with the *history/physical*, 45 (7.5%) were associated with *testing*, 0 (0%) were associated with *communication*, 47 (7.8%) were associated with *monitoring/follow-up* and 35 (5.8%) were *other pitfalls*. A breakdown of these diagnostic pitfalls per clinician type is given in online supplemental eTable 5 and online supplemental eFigure 4.

The five most prevalent diagnostic pitfalls were no specific diagnosis was ever made (*diagnosis/assessment* main category; n=271; 45.0%), endometriosis was often misdiagnosed as other diseases (*diagnosis/assessment* main category; n=123; 20.4%), counter-diagnosis cues overlooked (*diagnosis/assessment* main category; n=53; 8.8%), failure to appreciate limitations of a test result (*testing* main category; n=42; 7.0%) and failure to monitor, note or respond to evolving/continuing/persistent symptoms (*monitoring/follow-up* main category; n=30; 5.0%). A breakdown of the most prevalent diagnostic pitfalls taxonomy subcategories per clinician type is given in online supplemental eTable 6 and figure 3. See table 2 for specific illustrative examples for each specific diagnostic pitfalls taxonomy category, and patient exemplars are in online supplemental eTable 7.

Effect of diagnostic error on patients' health-related behaviour

Patients were not unaffected by these interactions and diagnostic delays. Patients either believed clinicians when they were reportedly told that their symptoms were 'normal', lost trust in clinicians for not acting on their symptoms, or even 'took breaks' from seeing clinicians entirely owing to the emotional and cognitive turmoil of these seemingly invalidation-laden interactions. Patients often reported not returning to the clinicians who invalidated and/or misdiagnosed them. Although this response is understandable, patients' not returning to the original clinicians who had reportedly invalidated and/or misdiagnosed them prevented clinicians' from being aware of their missed diagnosis or misdiagnosis, as they do not receive feedback from patients on their errors.[28]

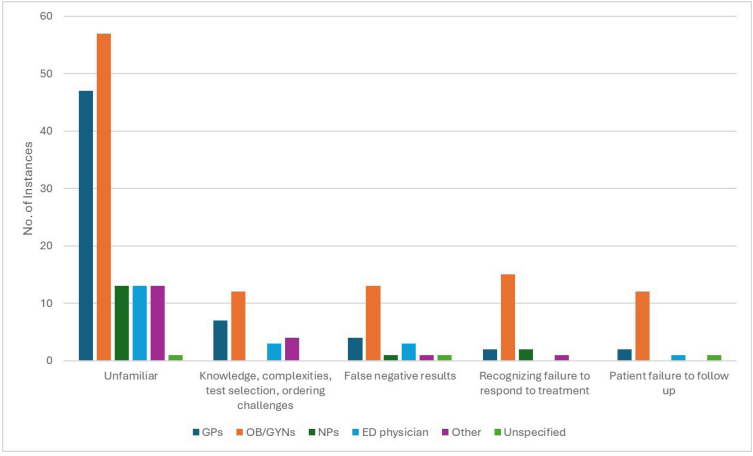

Figure 2 Five most prevalent Reliable Diagnosis Challenges Taxonomy subcategories by clinician type. ED, emergency department; GPs, general practitioners; NPs, nurse practitioners; OB/GYNs, obstetricians/gynaecologists.

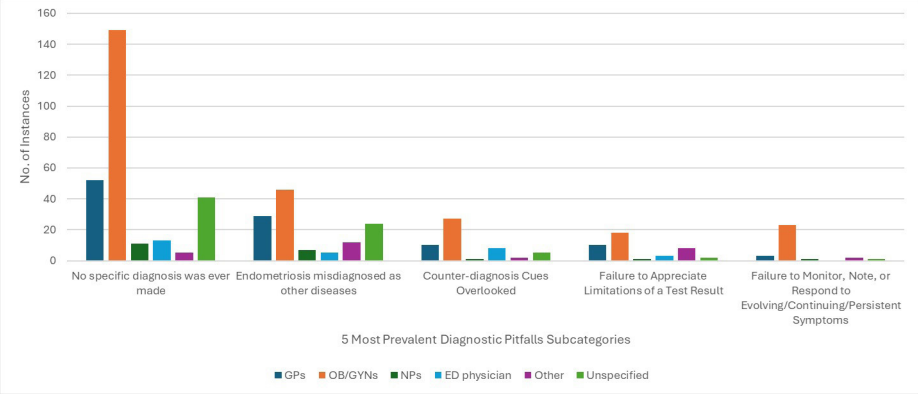

Figure 3 Five most prevalent diagnostic pitfalls taxonomy subcategories by clinician type. ED, emergency department; GPs, general practitioners; NPs, nurse practitioners; OB/GYNs, obstetricians/gynaecologists.

DISCUSSION

We systematically reviewed patient responses to a survey question to classify patient-reported breakdowns in the diagnostic process for patients with endometriosis by analysing qualitative data using taxonomies developed specifically for identifying opportunities for improvement in the diagnostic process. We provide a discussion of the results, which is organised by the order in which diagnosis typically unfolds (clinical history and interview, diagnostic testing, follow-up and referral).[2]

Reported breakdowns in the diagnostic information-gathering process

Clinical history and interview

The most prominent diagnostic error was the number of clinicians who had reportedly told patients that their symptoms were 'normal'—in most cases, normal menstruation or another benign process (eg, constipation, gas). This finding is consistent with a systematic review of qualitative studies and is reflected in the DEER Taxonomy subcategory of inaccurate/misinterpreted/overlooked critical piece of history data (*history* main category) and the diagnostic pitfall taxonomy subcategory of no diagnosis was ever made (*diagnosis/assessment* main category).[18] Furthermore, what predominated in the diagnostic pitfalls taxonomy subcategory of counter-diagnosis cues overlooked (*diagnosis/assessment* main category) was that the severity of symptoms and their interference in patients' lives were reportedly not appreciated by clinicians. Recognising symptom severity and impact is key in distinguishing normal menstruation from pathological processes such as endometriosis. Prior work demonstrates that clinicians report difficulty in distinguishing normal menstruation from pathological conditions such as endometriosis.[29 30] Patients reported their symptoms were often misattributed to ovarian cysts, irritable bowel syndrome, sexually transmitted diseases/resulting pelvic inflammatory disease, and mental health disorders. These specific misdiagnoses have been demonstrated in prior studies, in which nearly half (49.5%) of patients who reported having their endometriosis misdiagnosed initially were told their symptoms were due to a mental health disorder.[17 31 32]

Diagnostic testing

Failure to perform diagnostic testing

Given the frequency in patient responses that symptoms are misattributed to normal menstruation or another benign process (eg, constipation, gas) rather than to something potentially pathological, history-taking and diagnostic testing were foregone. Thus, the large number of cases where there was a reported failure/delay in ordering needed test(s) (*tests* main category) was not surprising. In many cases, no imaging (eg, ultrasonography) was ordered and/or performed. Though ultrasonography is not yet currently regarded as a definitive diagnostic test, the ordering and performance of an ultrasound can aid clinicians' assessment of the patient's symptoms and identify potentially pathological nature of patients' symptoms, or, at the least, rule out other potential diagnoses.[13] In other cases, OB/GYNs reportedly rejected patients' requests for diagnostic laparoscopy, even in cases in which patients had reportedly been suffering for many years.

Performance on non-discriminatory/non-helpful diagnostic testing

When symptoms were appraised by clinicians as potentially pathological, patients reported that clinicians often did order and/or perform various other diagnostic tests. Because these diagnostic testing methods are either not sensitive or specific (eg, blood tests) or are non-discriminatory in diagnosing endometriosis (eg, transvaginal ultrasonography), the results were often normal, potentially falsely leading clinicians to conclude that there was nothing wrong.[23] This finding is also consistent with the systematic review of qualitative studies in the context of endometriosis.[18]

Follow-up and referral

Often, patients reported being prescribed oral contraception with no response and often for much longer than the recommended 6 months by the American College of Obstetricians and Gynecologists for the management

Table 2 Illustrative examples of diagnostic pitfalls taxonomy categories as reported by patients (n=602)

Diagnostic pitfalls	Illustrative examples
Endometriosis often misdiagnosed as another disease	Endometriosis was often misdiagnosed as cysts, irritable bowel syndrome or sexually transmitted diseases/resulting pelvic inflammatory disease; endometriosis also often misdiagnosed as psychological
Misled by atypical presentation	Endometriosis located on rare sites for endometriosis, such as diaphragmatic and umbilical endometriosis, manifesting with atypical symptoms, such as chest pain and an umbilical mass, respectively
Rare diagnosis: failure to consider or know	N/A
Chronic disease presumed to account for new symptoms	New endometriosis symptoms attributed to pre-existing conditions of polycystic ovarian syndrome, coeliac disease, anxiety and depression
Counter-diagnosis cues overlooked (eg, red flags, things that do not fit not recognised)	Did not take into consideration certain symptoms and/or their symptoms' severity and interference with their life when they told patients that they were just experiencing normal menstruation or were misdiagnosed with another illness
Drug or environmental factor overlooked as cause of symptoms, or as cause of disease progression	N/A
No specific diagnosis was ever made	Told that nothing was wrong with them (symptoms were normal, experienced by all or most females or were part of being a female, were normal symptoms of a period, that periods are supposed to be painful, or that some females just experience really bad periods); symptoms attributed to poor coping with menstrual pain; told they were faking their symptoms, seeking attention and/or being dramatic; symptoms never investigated; symptoms due to being overweight; received treatment without a diagnosis
History/physical	
Nonspecific/vague symptom; hard to pinpoint diagnosis	N/A
Intermittent symptoms: overlooked because findings (eg, exam, lab, EKG) negative when seen	N/A
Failure to appreciate risk factor (or those at risk) for a given disease	Overlooked family history as a risk factor for endometriosis
Failure to appreciate limitations of the physical exam	Pelvic exam alone used to diagnose the patient; Pap smear alone used to diagnose the patient
Testing	
Failure to appreciate limitations of a test result(s)	False negative results from ultrasound, CT scan, X-ray, and MRI
Failure in follow-up of abnormal/critical result	Failure to communicate to patient that endometrioma was detected on ultrasound
Communication	
Communication failure with patient, including language barriers, health literacy, phone	N/A
Failure around communication and ordering of lab tests	N/A
Communication failure between physicians (eg, PCP-specialist, emergency department-PCP)	N/A
Monitoring/follow-up	
Failure to monitor, note, or respond to evolving/continuing/persistent symptoms	Patients tried on one contraceptive pill after another despite lack of response to it, clinicians reportedly failing to investigate via laparoscopy
Inadequate follow-up visits/referrals, especially in the presence of diagnostic uncertainty	Clinicians denying patients' requests to be referred to OB/GYNs or endometriosis specialists
Other	
Urgency of the clinical situation was not appreciated	At diagnosis, discovery of damage to multiple organs from endometriosis (eg, emergency oophorectomy and salpingectomy due to a 13 cm endometrioma that ruptured)

Continued

Table 2 Continued

Diagnostic pitfalls	Illustrative examples
Diagnostic findings were masked or misinterpreted due to an intervention or drug (eg, empiric treatment with oral or topical steroids, PPI, antibiotics, pain medications)	Suppression of symptoms due to contraceptive pill and/or pain medication
Problems with inappropriate or over-referral	Referral to fertility specialist when patients' chief concern was pain; referral to mental health specialist when patients' chief concern was pain

EKG, electrocardiogram; MRI, magnetic resonance imaging; PCP, primary care provider; PPI, proton pump inhibitors.

of their symptoms in lieu of having their symptoms investigated, which is reflected in the diagnostic pitfalls taxonomy subcategory of failure to monitor, note or respond to evolving/continuing/persistent symptoms (*monitoring/follow-up* main category of diagnostic pitfalls taxonomy).[33] This is a reported demonstration of clinicians' not engaging in diagnostic practices consistent with care guidelines. In the case of GPs, this included failure to refer patients to OB/GYNs. This finding is supported by focus group studies of GPs who have commented on not knowing when to refer patients.[30] In the case of misdiagnosis or delayed diagnosis by OB/GYNs, there were frequent reported failures to investigate patients' symptoms after a failed trial of oral contraception—a recommended first-line treatment for endometriosis.[13] Furthermore, there was a reported failure to refer patients to OB/GYNs with specialised training in endometriosis. It is the recommendation of the American College of Obstetricians and Gynecologists that for patients with dysmenorrhoea, for those who do not experience clinical improvement within 3–6 months after initiation of oral contraception, OB/GYNs should investigate for underlying pathological reasons for the pain, which includes endometriosis, including offering a diagnostic laparoscopy.[33] Thus, it is noteworthy that despite patients being symptomatic for years, being switched from one type of oral contraception to another without improvement, OB/GYNs reportedly failed to investigate symptoms beyond empiric treatment with oral contraception, a trial of which is recommended to be used for a maximum of 6 months.

Reported performance of OB/GYNs in endometriosis diagnosis

It is worth noting that OB/GYNs were the type of clinicians patients reported as making the largest number of diagnostic errors, accounting for a little over half 266 (55.9%) of all interactions in which a diagnostic error was reported. Although this in part reflects that patients sought care from OB/GYNs owing to the nature of their symptoms, this finding nevertheless warrants concern, given that specialists, more generally, should be better positioned to make a diagnosis in their specialty area, especially for a disease as prevalent as endometriosis.[34–37] Based on patients' descriptions, there appeared to be suboptimal knowledge of endometriosis among some OB/GYNs—how to recognise it, and how to establish the diagnosis—among the OB/GYNs the patients consulted. For example, endometriosis can only be diagnosed via laparoscopy and biopsy and cannot yet be diagnosed via transvaginal ultrasound.[13] Nevertheless, diagnostic testing was reportedly a contributor to diagnostic error among all three taxonomies. OB/GYNs were reportedly unaware that endometriosis can only be diagnosed via laparoscopy and biopsy, and cannot be definitively diagnosed via ultrasound.[13] As another example, there is a sevenfold familial risk of endometriosis, and among the coded instances of failure to appreciate family history as a risk factor for endometriosis (*history/physical* main category of diagnostic pitfalls taxonomy), all seven (100%) of the clinicians implicated in this pitfall were OB/GYNs.[38] As another example, among the number of instances in which patients reported suggesting to clinicians endometriosis as the candidate diagnosis but were reportedly dismissed by the clinician, 41 (77%) were OB/GYNs. Preliminary research indicates that OB/GYNs in the USA do not receive adequate education on and training in endometriosis.[39] There is no research to date that systematically assesses OB/GYNs' knowledge of endometriosis nor gauges OB/GYNs' experiences with diagnosing endometriosis in the USA; thus, there is a critical need for such research.

Reflecting on the coding taxonomies

Although we assert that the taxonomies are used complementarily, there was some overlap in the taxonomies in the coding process and thus the results. For example, this overlap is most overtly demonstrated for testing-related factors. For example, units of analysis were often double-coded or triple-coded related to testing (the DEER Taxonomy *tests* main category, the RDC Taxonomy *testing challenges* main category and the *testing* main category of the diagnostic pitfalls taxonomy). Thus, some redundancy was seen. Yet, in the DEER Taxonomy, the *tests* main category allows us to identify *where* in the diagnostic process an error occurred, which is complemented by the RDC Taxonomy

Nevertheless, the taxonomies facilitated the systematic identification of reasons for diagnostic error among clinicians as reported by patients with endometriosis (ie, our sample). Indeed, these taxonomies each contributed uniquely to our understanding of factors contributing

to diagnostic error. For example, one of the items in the DEER Taxonomy, failure/delay in considering the diagnosis, detected reported cases in which patients explicitly presented endometriosis as a differential diagnosis to their clinicians, which was subsequently reportedly denied. It is sobering to see that clinicians who were made aware of endometriosis as a diagnostic consideration by nevertheless deny endometriosis as a diagnostic possibility. One wonders what criteria, if any, these clinicians are using to diagnose (or not diagnose) these patients. As another example, the RDC Taxonomy was able to identify a unique, patient-focused factor that likely prolongs time to diagnosis: failure to return to the clinician or even 'taking breaks' from seeing clinicians. This result, then, reinforces the importance of patients and clinicians cultivating a strong relationship at the outset to improve diagnosis.

Thus, although there may be some overlap between items in the taxonomies, there appears sufficient uniqueness in detecting reasons for diagnostic error in our sample. During the inception of the diagnostic pitfalls taxonomy, the most recently developed taxonomy, these three taxonomies were also used complementarily with success.[25] For researchers looking to use these taxonomies in coding reasons for diagnostic error in their own research, perhaps a streamlined taxonomy could be implemented that would reduce some of the overlap.

Limitations

This study has several limitations. First, we used a convenience sample and recruitment that was primarily online which mentioned our interest in surveying patients with errors and/or experiencing symptom invalidation. This intentionally enriches as well as biases the sample to selectively include more patients who experienced delays or errors. Nonetheless, the findings mirror those from less selective cohorts with this diagnosis.[21] Our findings may not be applicable to all patients with endometriosis, especially given the demographic profile of patients—well educated, non-Hispanic white. Second, for some patients, there was uncertainty when coding responses owing to insufficient data in the open-ended responses, as the prompt did not specifically inquire about diagnostic errors, given the secondary analytic nature of this study. Thus, the specific codes that predominated may indeed reflect clinician actions that are correspondingly more dismissive, biasing our results. In our coding process, we only checked off when there was explicit affirmative evidence of one of these things going wrong in the diagnostic process, though we must also recognise that there were items that were not included in patients' responses that may be, for example, in the medical record that would not have been picked up to code as positive taxonomy items. Furthermore, the prompt asked specifically about one interaction, rather than patients' whole diagnostic journey. Nevertheless, a portion of patients reported interactions with more than one clinician. Because this prompt focused on patient-clinician interactions, patient-associated factors and system-level factors are likely under-represented, precluding a more complete picture of diagnostic error in endometriosis. Although not discussed in patient responses to the prompt, this factor likely contributed to delays in *seeking care* which was, on average, 4.1 years. That some patients described one versus multiple interactions made counting frequencies difficult. The coding was predominantly done by a single individual (ie, the first author) although any uncertainties were reconciled by discussion with the clinician co-investigator. No data were collected from clinicians' perspectives, nor paired chart reviews, research that is generally lacking in the field. Finally, the sample comprised entirely patients who self-identified as having endometriosis; we were unable to confirm patients' self-identified diagnosis.

CONCLUSION

Despite the high prevalence of endometriosis, the cohort in our study reported clinician failure to recognise and/or definitively diagnose their endometriosis symptoms. This produced prolonged diagnostic delays, often several years. By systematically analysing these diagnostic errors for a large sample of patients with endometriosis, findings suggest that clinicians often erred in their appraisal of symptoms, labelling them as normal menstruation and drawing incorrect conclusions from results of non-discriminatory diagnostic testing. These shortcomings in achieving timely and optimal diagnosis were reported for both general practitioners and OB/GYN generalists and suggest the need for systematically assessing the performance of front-line clinicians and dispelling misinformation. These findings can help patients, clinicians and healthcare organisations design and implement education efforts and safety nets for the anticipation, prevention and/or mitigation of such errors.

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