Scientists as Patients and Patients as Scientists

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Disclosure

I have no conflict of interest to report.
Hi, I’m Noémie

I like computers and medicine.
Hi, I’m Noémie

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Reddit AMA:
Menstrual Health and Endometriosis

Hosted by r/TwoXChromosomes
Thursday, February 28, 11am EST

Women Discuss The Invisible Pain Of Endometriosis
Endometriosis – Ask a gynecologist

• Endometrial-like cells outside the uterus
• Diagnosis is done through surgery
• Symptoms include dysmenorrhea and infertility
• Treatments are surgical and hormonal
Endometriosis – Ask an epidemiologist

• 1 in 10 women in reproductive age
• Delayed diagnosis of 4-17 years
• No established risk factors
• Increased risk for ovarian cancer, heart disease
• High morbidity, with loss of productivity
  • Average 10 hours / week
Endometriosis – Ask a clinical researcher

• 4 surgical disease stages/ 3 histological phenotypes established
• No known biomarkers to diagnose or monitor progression
• No understanding of which treatment will work for whom
• No cure
• SNPs identified through GWAS but with low explanatory power

“a riddle wrapped in a mystery inside an enigma”
-Emory Wilson, MD
Endometriosis: an enigmatic disease with many faces

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ABSTRACT

Endometriosis is a benign disease defined by the presence of the endometrial glands and stroma outside of the uterus, both at pelvic and extra pelvic sites. We are reporting 4 unusual cases of endometriosis that are unique on their own due to site of endometriosis, presenting symptoms, complications and treatment. Endometriosis was present at post caesarean scar site in one case, at vault site in another case, in bladder in third case and in the last case it caused fracture pelvis with haemorrhage, hydronephrosis and primary infertility. All four patients have been treated.

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Call to Action
Clinical diagnosis of endometriosis: a call to action
Sanjay K. Agarwal MD 1, Charles Chapron MD 2, Linda C. Giudice MD, PhD 3, Marc R. Lauffer MD 4, Nicholas Leyland MD 5, Stacey A. Misermer ScD 6, Sukhbir S. Singh MD 7, Hugh S. Taylor MD 8, 9

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Endometriosis: where are we and where are we going?
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(A D Greene and S A Lang contributed equally to this work)

Abstract

Endometriosis currently affects ~3.5 million reproductive-aged women in the U.S. with symptoms such as painful periods (dysmenorrhea), chronic pelvic pain, pain with intercourse (dyspareunia), and infertility. It is defined as the presence of endometrial tissue outside the uterine cavity and is found predominately attached to sites within the peritoneal cavity. Diagnosis for endometriosis is solely made through surgery as no consistent biomarkers for disease diagnosis exist. There is no cure for endometriosis and treatments only target symptoms and not the underlying mechanism(s) of disease. The nature of individual predisposing factors or inherent defects in the endometrium, immune system, and/or peritoneal cavity of women with endometriosis remains unclear. The literature over the last 5 years (2010-2015) has advanced our critical knowledge related to hormones, hormone receptors, immune dysregulation, hormonal treatments, and the transformation of endometriosis to ovarian cancer. In this review, we cover the aforementioned topics with the goal of providing the reader an overview and related references for further study to highlight the progress made in endometriosis research, while concluding with critical areas of endometriosis research that are urgently needed.
Endometriosis – Ask me

• Symptomatic for 30 years
• Hormonal treatments for 20 years, pain killers
• 7 endo-related surgeries + 1 complication follow-up surgery
  • Endometriosis lesions found on 8 organs (left alone, resected, or removed)
• Misdiagnosed with 3 cancers, IBD, 2 auto-immune diseases
Endometriosis – Ask the patients
Endometriosis – Ask the patients

“It has ruled (and ruined) my life since I was 13. I’m 30 now and feel like I’ve had no life.”
Endometriosis – Ask the patients

“It has ruled (and ruined) my life since I was 13. I’m 30 now and feel like I’ve had no life.”

“At 32, I have exhausted the treatments available. Endo has taken my social life, sex life, and I have to struggle to work. Now it has taken my ability to have children… I feel like endo has taken my identity. I grieve for the woman I would have been without synthetic hormones and medication, would I have been a different person without endo? Unfortunately I’ll never get to know who the ‘real me’ ever was. That is my biggest loss.”
Endometriosis – Ask the patients

“It has ruled (and ruined) my life since I was 13. I’m 30 now and feel like I’ve had no life.”

“I have been suffering since I was about 18 and I am going to be 48. I often wonder what I could have accomplished if I not had this monster disease. I have sex life, and I have to struggle to work. Now it has taken my ability to have children... I feel like endo has taken my identity. I grieve for the woman I would have been without synthetic hormones and medication, would I have been a different person without endo? Unfortunately I’ll never get to know who the ‘real me’ ever was. That is my biggest loss.”
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“I have been suffering since I was about 18 and I am going to be 48. I often wonder what I could have accomplished if I not had this monster disease. I lost my sex life, and I have to struggle to work. Now it has taken my ability to have children... I feel like endo has taken my identity. I grieve for the woman I would have been without synthetic hormones and medication, would I have been a different person without endo? Unfortunately I’ll never get to know who the ‘real me’ ever was. That is my biggest loss.”
Endometriosis – Ask the patients

#THISISENDOMETRIOSIS

@georgiewileman

I N V I S I B L E

ILLNESS

WHAT YOU
SEE

WHAT I
FEEL

no
mobility
problems
radiating
pain
no pain

migraines

no pain

brain fog

widespread
pain

fatigue

shooting
pain

hand

cramp

back

pain

injury

back pain

Stiffness

Joint pain

foot pain

lazy

if I can't
do something
The problem

• There is a disconnect between the way patients experience the disease and its current scientific characterization
  • Proposed phenotypes do not correlate with symptoms and their severity
  • Current characterization ignores many of the symptoms and their temporal trajectories

• Impacts detection, monitoring, and research

• Frustrating to patients
  • Loss of trust in doctors and scientists
• How do I get better?
  • How do I stay functional? Active member of my family, community?
  • How do I do it without quitting my job and breaking the bank?
• Is my daughter at risk? → is that endo?
• What will happen to me?
• How will I know if something *really* bad happens to me?

• Detection / Phenotyping
• Decision making
• Trajectory prediction
(Patient | Scientist) \(\rightarrow\) Patient Scientist

• I am
  • Keeping my endo as private as possible (stigma of invisible illness)
  • Keeping my personal and professional life compartmentalized

• I should use my data-science skills to contribute endo research
  • The data I have access to (EHRs, claims) agrees with the gyne and epi views
  • There are patient narratives online, but is this enough to advance knowledge of disease?
  • Are the questions I asked myself as a patient any relevant to current scientific questions?

• I want to do research in endometriosis
  • “Why? Do you have endo?”
  • “You might be too emotionally invested to do good research”
  • “Join the club”
• Interviews (n=3)
• Focus groups (n=27)
• Online surveys (n=741)
• Content analysis of online endometriosis community (1,500 posts)

Mental model of disease

Researcher-based

Patient-based
Designing in the Dark: Eliciting Self-Tracking Dimensions for Understanding Enigmatic Disease

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ABSTRACT
The design of personal health informatics tools has traditionally been explored in self-monitoring and behavior change. There is an unmet opportunity to leverage self-tracking of individuals and study diseases and health conditions to learn patterns across groups. An open research question, however, is how to design engaging self-tracking tools that also facilitate learning at scale. Furthermore, for conditions that are not well understood, a critical question is how to design such tools when it is unclear which data types are relevant to the disease. We outline the process of identifying design requirements for self-tracking endometriosis, a highly enigmatic and prevalent disease, through interviews (N=3), focus groups (N=27), surveys (N=741), and content analysis of an online endometriosis community (1500 posts, N=153 posters) and show value in People with chronic conditions, who are often faced with a complex set of decisions and environments to navigate, have additional incentives to understand and manage their condition, and thus engage in self-tracking.

Self-tracking systems that “help people collect personally relevant information for the purpose of self-reflection and gaining self-knowledge” are part of personal informatics, as defined by Li and colleagues [59]. Within the health domain, personal informatics tools have traditionally focused on self-monitoring for individuals to gain health-related self-knowledge or achieve a health-related goal [26,54]. In fact, designs of such tools have been proposed and evaluated for many chronic diseases, including diabetes [5,41,69,80,91], COPD [12,102], cardiovascular diseases [4,96], and Parkinson’s [11,72,77].
Citizen Endo (citizenendo.org)

• Community
• Meetups, “challenges”
• Email > Instagram > Facebook & Twitter
• Patient advocacy
Citizen Endo

• A research self-tracking app
Learning from self-tracked data

Phenotyping Endometriosis through Mixed Membership Models of Self-Tracking Data

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Abstract

We investigate the use of self-tracking data and unsupervised mixed-membership models to phenotype endometriosis. Endometriosis is a systemic, chronic condition of women in reproductive age and, at the same time, a highly enigmatic condition with no known biomarkers to monitor its progression and no established staging. We leverage data collected through a self-tracking app in an observational research study of over 2,800 women with endometriosis tracking their condition over a year and a half (456,900 observations overall). We extend a classical mixed-membership model to accommodate the idiosyncrasies of the data at hand (i.e., the multimodality of the tracked variables). Our experiments show that our approach identifies potential subtypes that are robust in terms of biases of self-tracked data (e.g.,...
Learning from self-tracked data

Participants ordered by phenotype assignments

Participants ordered by number of days tracked

Participants ordered by number of observations tracked
Patient questions beyond phenotyping

- Role of menstruation and hormones?
- Role of environment?
- Is there a connection between endo and X?
- How do I know whether I am getting sicker?
- How do I help my care team take care of me?

“I cannot tell what’s possible scientifically, so it’s hard for me to tell whether my questions make sense, but I have so many”
NARRATIVE MEDICINE
A Model for Empathy, Reflection, Profession, and Trust

Rita Charon, MD, PHD

The effective practice of medicine requires narrative competence, that is, the ability to acknowledge, absolve, interpret, and act on the stories and plights of others. Medicine practiced with narrative competence, called narrative medicine, is proposed as a model for humane and effective medical practice. Adopting methods such as close reading of literature and reflective writing allows narrative medicine to examine and illuminate of medicine’s central narrative situations: physician and patient, physician and self, physician and colleagues, and physicians and society. With narrativeme, physicians can reach and join their patients in illness, recognize their own personal journeys through medicine, acknowledge kinship with and duties toward other health care professionals, and inaugurate consequent discourses with the public about health care. By bridging the divides that separate physicians from patients, themselves, colleagues, and society, narrative medicine offers fresh opportunities for respectful, empathic, and nourishing medical care.

The BMJ has patient stories for over 20 years, and they have long been a dimension to our content and our identity. More than just the previous BMA Stories, but written with an even more compelling voice, the stories will give us a chance to link our stories of illness and care. For the patient, narrative medicine, supported by patients, can spread to other professionals. Our strategy was to produce with an international patient advisory group and content to be shared with them. The timely exchange with and between patients and medical professionals, patient-centered narratives of illness and care that make us partners and allows us to disseminate decisions across the journal. Patient narratives are often among the first to comment on emerging and promising therapies and their impact, which is often unconsidered or poorly understood by health professionals. We are now exploring the principles of narrative medicine for clinical practice, policy, and medical situations.

Although we’ve nationalized the analysis of communication, we recognize that sharing numbers is not enough. It is the quality and quality of the stories that matters. For our editorial content, we have established a platform for cooperation and exchange of patients’ narratives. Correspondence and review of educational articles by patients and others can provide a wider understanding of their interest and how they impact the patient. We are now exploring the principles of narrative medicine for clinical practice, policy, and medical situations.

The BMJ has a long tradition of patient participation in our content. A patient’s narrative is a platform for sharing our stories of illness and care with others. The BMJ has made strides in this area and uses it as a tool for training and education. The narrative medicine strategy has been a powerful tool for patient and policy programming, supporting narrative medicine in clinical practice, policy, and medical situations.

Better together: patient partnership in medical journals
The BMJ’s experience can be springboard for others
Tessa Richards, senior editor; patient partnership, Sarah Aich, senior research editor, Amy Price, patient editor; Fiona Godlee, editor in chief
The BMJ, London, UK

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• Patients and scientists each contribute to advancing medicine in complementary ways

• Go beyond existing datasets and convenient questions that can be answered just because of availability of current datasets

• Partner with patients
  • Observe them, learn from their experiences, but also their questions and ideas
  • Develop a common language
  • Feed your results back to patients
  • Iterate
Thank you!