

'Delicate diagnosis':

avoiding harms in difficult, disputed, and desired diagnoses

Medical diagnoses can be subject to ambiguity, flux, subjectivity, and inherent uncertainty. This is particularly true in primary care, where many reported symptoms do not conform to a clear diagnosis. Thresholds around normality are often unclear.

Symptoms are commonly experienced and described as an 'iceberg'. Over a third of otherwise well people without a chronic condition have felt tired or run down, or had a headache in the previous 2 weeks, and over a quarter have had back or joint pain. Distinguishing conditions that would benefit from diagnosis and earlier intervention from those that are temporary, self-limiting, and prone to harmful medicalisation, remains challenging.

While overdiagnosis is a diagnosis of a condition that, if unrecognised, would not cause symptoms or harm, contested diagnoses are symptoms given a diagnosis, but where the diagnosis, as an explanation for them, is controversial. Opponents consider contested diagnoses 'wrong' not because of an error in history taking, clinical examination, or interpretation of a test result (which would risk misdiagnosis), but because the diagnosis itself, for example, chronic Lyme disease or multiple chemical sensitivity, is invalid.

Contested diagnosis is bordered by 'pseudoscience', for example, adrenal fatigue, leaky gut syndrome, and chronic candidiasis, which attempt to explain symptoms with 'science' that is demonstrably incorrect.

Many contested diagnoses relate to symptoms that are not clear-cut, meaning that some people are given a diagnosis that does not confer benefit. Others are primarily unexplained 'yet'. For example, Gulf War syndrome was treated as a contested illness but is now recognised as caused by exposure to sarin gas. Other recognised conditions are prone to a variety of influences such that diagnosis risks causing harm, through a variable mixture of misdiagnosis, overdiagnosis, pseudoscience, or overtreatment.

A framework for considering some conditions as 'delicate diagnoses' may help to recognise and reduce the harms of medicine in these circumstances.

DELICATE DIAGNOSIS

A variety of models have been used to describe and explain how diagnoses are

reached by doctors. Heuristics – rules-of-thumb – discounts less important information to pragmatically reach likely probabilities, while responding to environmental circumstances. Bayes' rule means that small pieces of information can have a profound impact, with prior probabilities of disease rapidly shaping possibilities.

GPs work mainly from 'mindlines', 'collectively reinforced, internalised, tacit guidelines ... informed by brief reading, but mainly by their interactions with each other and with opinion leaders, patients, and pharmaceutical representatives'. Similar heuristic styles may be true for patients. This gives rise to the potential for bias, and the associated harms of poor-value care, overdiagnosis, and overtreatment.

THE PART DOCTORS AND GOVERNMENT PLAY

Finding good-quality information is often difficult. For example, searching for 'Do I have ADHD?' on Google brings up, in the first few hits, a trivia quiz site with an unvalidated questionnaire, a questionnaire hosted by a site sponsored by a manufacturer of drugs for ADHD, and only then, NHS information. The top hits for information about 'cow's milk protein allergy' includes baby milk manufacturers and patient organisations sponsored by them. Campaigns regarding prostate cancer have been run by men's organisations and consultants in private practice.

Nor have GPs in the UK been able to approach certain diagnoses neutrally. They have been previously financially incentivised under their contract to seek to make diagnoses 'early' that they would not otherwise have considered clinically useful. Despite continuity of care being desired by patients, it is in decline. Patients seek internet advice for ease, but doctors for professional skill – yet this may not be readily available given the stress in primary care.

In 2005, the Health Select Committee (HSC) review of the pharmaceutical industry

argued that *'The Department of Health has not only to promote the interests of the pharmaceutical industry but also the health of the public and the effectiveness of the NHS ... The Secretary of State for Health cannot serve two masters.'*

However, in 2013, the government launched Academic Health Science Networks (AHSN), seeking to increase the speed of the adoption of innovations within the NHS and 'an entirely new relationship with industry based on partnership, not just transactions'. These delivered a programme – screening for atrial fibrillation – that the UK's own independent committee on screening has recommended against.

In this context, full of competing interests, challenges in finding definitive evidence, and with gaps in NHS care, some diagnoses, and the circumstances in which they are likely to occur, may be considered 'delicate', meriting special care to navigate them. They feature many overlapping, fuzzy margins, unsettled science, and cultural differences, together with subjectivity of symptoms and risks of transactional elements (for example, payment to access a sought-after condition and associated sequelae).

These features – not intended to be exhaustive or definitive – may be especially important when considering diagnosis without the context of experienced heuristics or multiple means of sense checking via mindlines – or when these are systematically biased or based on poor information. The combination of sensitivity of area, expectations, and the difficulty of achieving certainty make them particularly prone to influence, including seeking diagnosis, and the threshold at which it is made.

A SHARED UNDERSTANDING BETWEEN DIAGNOSTICIANS, CITIZENS, AND PATIENTS

It may not be possible, or desirable, to make 'delicate' diagnoses more robust. Instead, a tolerance of uncertainty, and willingness to adopt a questioning, tentative, or provisional

"Tolerance of uncertainty and willingness to adopt tentative or provisional diagnosis may be an appropriate means of balancing risk."

Box 1. Challenges of a 'delicate diagnosis'

Type	Why	Features	Strategies
Difficult	<ul style="list-style-type: none"> Subjective, fluctuating symptoms may be variations of normal or common to multiple other conditions; and diagnosis itself might be sensitive 	<ul style="list-style-type: none"> Online questionnaires; poor-quality evidence for interventions; lack of control groups; low pretest probabilities; and other more likely explanation 	<ul style="list-style-type: none"> Use of time as a diagnostic tool; express chances of benefiting from diagnosis in absolute (not relative) terms; honesty about the risk of inaccuracy and hazards of diagnosis; use of/generate data about control groups; and computer coding that recognises uncertainty
Disputed	<ul style="list-style-type: none"> No broad medical consensus or strong challenge to diagnostic validity exists 	<ul style="list-style-type: none"> Disputes may arise between: lobby groups; professionals; industry; guideline authors; activists; regulators; private sector; insurance companies; and the NHS and its staff 	<ul style="list-style-type: none"> Clear statements of conflicts of interest and mitigation strategies, including what conflicts are not manageable and should lead to exclusion; agreed, official 'cross-party' explanations recommending independent advice; and efforts to achieve a multiparty consensus
Desired	<ul style="list-style-type: none"> Single issue pressure groups/clinics seeking increased diagnosis rates, awareness campaigns, contractual targets, and media campaigns; patient preference for rapid diagnosis; and high expectations of value of diagnosis and consequent intervention 	<ul style="list-style-type: none"> Strong media presence; industry influence via third sector; charitable or pressure organisations; few/no downsides presented; claims of underdiagnosis; and social pressure put on patients/citizens 	<ul style="list-style-type: none"> Seek independent information that balances benefits and risks of diagnosis; consider that 'single issue' diagnostic routes create a bias within themselves; and advocate for holistic assessment considering wider possibilities for cause of symptoms

diagnosis may be appropriate. This has to be set against the potential for harm where there may be value in diagnosis and consequent treatment. However, urgent intervention in these cases is rarely needed, and rapid diagnosis has to be set against the harms of 'diagnostic overshadowing, low-value, and harmful medicine, and the burden of 'patienthood'.

When the prevailing environment reflects a bias, work may be required to achieve and maintain a more neutral equipoise (Box 1). Computer coding systems may reflect a potential or uncertain diagnosis, and may require expanding to reflect uncertainty. Nor should the value of diagnosis be underplayed: the ability to name a disorder gives the ability to develop robust definitions, fairly test, and develop effective interventions.

The challenge for clinicians and patients is to find benefits while avoiding harms. This may not be easy for either party, especially given the attractiveness of certainty and privileges associated with a particular diagnosis. Minimising harm and maximising benefit requires partnership between doctor and patient, especially in an environment replete with vested interests.

CONCLUSION

As medicine shifts in the era of COVID-19, new challenges are likely to impact on

how and why doctors and patients seek diagnosis, and the culture in which they do it. Continuity and relationship-based care are valued but are becoming less common. A panoply of environmental pressures, some visible and some not, are prevalent. New and emerging information can be difficult to critically appraise and use, often because of biases and misinformation.

A framework to consider some diagnoses as 'delicate' allows for systematic identification and analysis of the influences surrounding them. This may help organise appropriate mitigations against bias, help patients and doctors avoid harm, and inform research and policy.

KEY POINTS

- Some diagnoses are particularly prone to ambiguity, overdiagnosis, overtreatment, and associated harm;
- systematic factors include bias, conflicts of interest, and variability in information quality for patients and clinicians;
- considering certain diagnoses as 'delicate' allows identification and potential mitigation of these features; and
- tolerance of uncertainty and willingness to adopt tentative or provisional diagnosis may be an appropriate means of balancing risks.

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