The changing nature of diagnosis

Implications for policy, practice and patients

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Authors

Tanya Brigden, Jamie Hearing, Heather Turner

Correspondence to

PHG Foundation can be contacted at intelligence@phgfoundation.org

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1. Introduction

Advances in medical imaging, molecular testing (including genomics), data-driven analysis and artificial intelligence offer new opportunities for diagnosis. These innovations expand possibilities for investigating the causes of ill health, describing and differentiating between diseases, and detecting biological signs of disease before the onset of symptoms. This has implications for the scope, timing, precision and degree of certainty associated with diagnosis, raising issues of growing relevance to patients, healthcare professionals and health policy makers.

Working with Fellow, Dr Zoë Fritz[1], the PHG Foundation convened a multidisciplinary group of clinicians and academics, with expertise across different medical specialities, as well as in public health, biomedical science, ethics, philosophy and sociology, to find out how diagnosis is changing in response to technological innovation and to identify priorities for future research, policy and practice.

Through an initial roundtable discussion, we aimed to:

- Understand how the practice of diagnosis is changing.
- Identify or anticipate challenges for norms of practice, clinical governance and policy in this area.
- Explore the need for different actors in the health system to respond.

1 Dr Zoë Fritz is Associate Professor in Medical Ethics and Ageing based in THIS Institute and a Consultant Physician in Acute Medicine at Addenbrooke's Hospital. Her research includes work on the clinical and ethical implications of diagnostic uncertainty.

2. How is diagnosis changing in different clinical contexts?

The practice of diagnosis involves different clinical specialities whose role depends on their clinical context. We heard from clinicians who work in contrasting settings about how diagnosis is changing in their practice.

2.1. Chronic disease: Parkinson's disease

Biomedical science has been uncovering the drivers of chronic disease with power to radically change diagnosis. Parkinson's disease epitomises this phenomenon with recent and rapid changes to how clinicians are approaching diagnosis. Parkinson's disease is diagnosed based on clinical presentation with some now advocating detection of α-synuclein pathology to establish a definitive diagnosis[2]. Diagnosis is often at a later stage and current treatments manage symptoms and do not represent a cure. Earlier detection of the presence of abnormal neuronal α-synuclein is possible before the onset of symptoms. Nevertheless, diagnosis is ahead of treatment for Parkinson's disease and this raises new questions that weren't present before, such as, do patients want a diagnosis when there is no evidence-based treatment to offer?

2.2. Primary care and the limitations facing GPs

Primary care is faced with a wide range of patients with symptoms that are often undifferentiated. Clinical history is central to decision making, but GP appointments are short, leaving limited time to address patient needs. The role of a GP in diagnosis is to triage the right course of action – watch and wait, onward referral or limited investigations. Limitations in terms of access to diagnostic tools – for example, genetic testing, blood tests or imaging – restrict the options available to GPs, with geographic variation in access. Digital tools are enabling new models of service delivery, including remote triage and prioritisation. This can improve clinical responsiveness but may introduce, or compound existing, inequities.

2.3. Emergency medicine

As in primary care, patients present with variable – yet acute – needs in emergency medicine. A large team is involved in decision making. Many patients arrive with complex needs and a diagnosis of exclusion, or a partial diagnosis, is a common outcome. This diagnosis is

2 Simuni T, Chahine LM, Poston K, Brumm M, Buracchio T, Campbell M, et al. A biological definition of neuronal α-synuclein disease: towards an integrated staging system for research. The Lancet Neurology. 2024 Feb 1;23(2):178–90. DOI: 10.1016/S1474-4422(23)00405-2

important for managing patients' acute needs, but there is limited scrutiny of links between diagnosis and outcomes following discharge from A&E. In contrast, we heard that regular debriefing is undertaken for air ambulance medicine. Cutting edge diagnostic tools have been implemented in air ambulances, but patients often have a very poor prognosis. Regular meetings with pathologists allow clinicians to review patient outcomes, to understand what they might have missed, and to undertake ongoing learning and development in this high-stakes setting.

2.4. Genomics and a molecular diagnosis

Rapid technological advances in genomic medicine have unlocked genetic diagnoses for rare disease patients. The purpose of a genetic test is to identify a unifying diagnosis that explains many, if not all, symptoms experienced by the patient.

A genetic diagnosis can be life changing, alleviating the sense of guilt often felt by family members/ parents, and enabling tailored insights into prognosis, surveillance and family testing. Increasingly, genomics is being considered for screening, particularly to replace traditional phenotype-led newborn screening programmes.

The proposition is greater flexibility to screen for a wider range of conditions. This raises ethical and scientific challenges. Evidence is growing that penetrance, the likelihood of a genetic variant presenting as disease, may have been overestimated because most estimates are from families with known history of disease. How predictive is genetic testing and how do we communicate this uncertainty? Currently, we do not have the language to describe some of these nuances to patients.

2.5. Summary

Across clinical contexts, clinicians seek to provide clear guidance for patients but, depending on the context, challenges exist in communicating uncertainty in diagnosis to patients. Technological advances are enabling and redefining how clinicians view the role of diagnosis in treatment decisions. In some contexts, resource and capacity limitations mean that patients do not receive a final diagnosis, but this distinction may not always be clearly communicated.

3. Challenges arising from the changing nature of diagnosis

Challenges arise, which can be brought under five key themes: the use of diagnostic tests, the clinical practice of diagnosis, informing and supporting patients during the diagnostic process, constraints and complexity in the health system, and the effect of diagnostic technologies on equity of access and performance.

3.1. Diagnostic tests

Diagnostic tests are used in different circumstances to rule in, rule out and confirm diagnoses, as well as to inform treatment management. However, these purposes do not appear to be clearly differentiated and there is limited data collection to understand how test ordering relates to clinical judgement. Current use of diagnostics raises a number of considerations that require further attention:

- Defensive medicine describes clinical practice that is driven by a desire to reduce the risk of malpractice lawsuits, for example by ordering unnecessary tests, which could come at the expense of maximising the benefit to patients.
- Diagnostic test stewardship defines the circumstances of when tests should be used, to improve patient outcomes, and the processes to control use when considering optimal use of resources.
- An increase in misdiagnosis can arise from broader use of diagnostics, for example in larger populations, for pre-clinical disease, or as a screening tool.
- Harms can arise for patients, such as from overtreatment, as well as additional clinical workload, from overuse of diagnostics.

High-quality evidence collected on the use of tests and clinical outcomes could inform critical questions about the use of diagnostics. However, evidence collected on the use of diagnostics across the health service is currently variable. Additionally, while extensive data may be collected before a test is implemented (e.g. the ACCE framework for genetic testing, or UK National Screening Committee guidance for screening tests), the evidence on the use of diagnostic tests is poor, particularly when comparing use across regions.

3.2. Clinical practice

Healthcare professionals have different roles in diagnosis, collecting clinical history, ordering diagnostic tests and directing patients along clinical pathways. The term diagnosis is used in different ways and has different meanings among patients and clinicians. A diagnosis places a label on individuals with consequences for them and this functional description has implications in medicine (i.e. treatment) and socially (to access support and as part of social identity). These differences raise questions about whether more specific language or terminology is needed to communicate the nuance of this process to patients.

Tests provide a useful example to frame this challenge. The same test may be used for screening as well as diagnosis, but context is important, and different populations may be targeted depending on the purpose of a test. A transition towards a preventative model of healthcare delivery suggests a move towards more screening and early detection of disease. Populations targeted for these interventions (typically asymptomatic) represent a broader section of the population. Decision makers must balance achieving health and addressing illness against risks of overmedicalisation, for example from unnecessary screening or interventions. This introduces uncertainty for clinicians when communicating risk to patients.

Artificial intelligence offers the opportunity to automate and improve consistency across the regular and repetitive work delivered by health services. This is particularly valuable across a national health service, where there is the opportunity to deploy tools to support more equitable service delivery. Digital tools can reduce variation in clinical judgement and mitigate against differences in experience known to result in variation in clinical outcomes. Deployment of digital tools, if done well, could help clinicians to dedicate more time to patients throughout the diagnostic process.

3.3. Informing and supporting patients

Patients have always provided and received information during the diagnostic process but the norms that guide this are subject to change as technology and clinical practice evolve. Among clinicians, there is a lack of clarity around the optimum amount of information to communicate to patients. There is clear need for clinical skills in communicating risk and uncertainty to patients, amid growing amounts of information of uncertain clinical relevance and widening access to this.

Autonomy is a core principle within contemporary medicine, supported in part by the exchange of knowledge between clinicians and patients. However, there is concern about the provision of uncertain diagnostic information without due attention to its context or potential harms. Clinicians may decide to be transparent about the diagnostic process but avoid disclosing all information in order to manage uncertainties and support patients in the way they consider most appropriate. This could be interpreted as paternalistic, for example if it excludes patients from important decisions about their care. This indicates the importance of trust, specifically trust in the discretion of the clinician, in determining if it is appropriate to withhold information.

As both clinicians and patients acquire greater access to information, particularly through the use of digital tools, it may be challenging for clinicians to determine what information should be communicated to patients, and when. Clinicians may be required to support patients

to understand types of information they have not previously disclosed, either because this was not possible or it was deemed unhelpful. Much of this information may be of uncertain significance in the immediate term and will require shared decision-making by clinicians and patients to determine if it is clinically appropriate to pursue or pause further investigations to establish a diagnosis. Clinical action is often perceived more favourably by patients and the line between clinical discretion and managing finite health resources is often unclear.

3.4. Constraints and complexity in the health system

The diagnostic process is embedded in clinical pathways within the health system. The structure of the health system shapes the decisions made by clinicians and patients during the diagnostic process. This system is structured by policies and regulations, as well as influenced by professional bodies, patients and industry.

Resource constraints exist across the health service in terms of time, resource and capacity. A preventative model of health delivery prioritises screening and earlier diagnosis as the solution to the population health crisis, however, in the interim, additional capacity is needed to deliver services through a transition. Test stewardship has a role here to ensure that tests are used to maximise their benefit, while being mindful of resources used for service delivery.

Ageing and increasing multimorbidity is seen as a critical challenge across the health system. Patients present similarly and, yet, with complex health needs arising from multimorbidity and polypharmacy. Current health services address acute illness and not health, despite optimal health outcomes likely requiring a holistic approach. The specialist nature of health services may compound this problem, with clinicians taking responsibility for parts of, but not the complete, diagnosis. Complex care pathways with multi-specialist involvement increases complexity for healthcare professionals working in the health system to understand their role in diagnosis.

3.5. How do inequities arise?

The implementation of new diagnostic technologies can pose challenges for ensuring the health benefits accrued are distributed equitably across populations. There is a need to consider how innovations can be distributed fairly within a health system, for example by prioritising their deployment in areas associated with more urgent need and by avoiding over-concentration in areas which benefit from high levels of research and development.

Without evidence that diagnostic technologies can work effectively in a range of settings, patchy implementation risks widening health inequalities. Where more precise diagnosis is possible, this may depend on hyper-specialist knowledge held by a small number of individuals, who are likely to only be accessible to patients via specific, multi-staged clinical pathways. Facilitating equitable access in this context introduces additional challenges.

Technological innovation can also disrupt the traditional routes through which individuals access care. Increasingly, patients can access information which may be relevant to diagnosis via third parties, such as through direct-to-consumer testing, and this may offer a route to diagnosis that is not universally available. This risks exacerbating existing inequalities in health knowledge across the population, which could in turn affect who gains access to and takes up health services. In the context of diagnosis, this could mean the difference between an early and late diagnosis.

4. Future of diagnosis

As this roundtable confirmed, rapid technological advancements are impacting the practice of diagnosis across clinical domains. They are giving rise to profound challenges in ensuring both patients and healthcare professionals are equipped to navigate new models of predictive and preventative testing, and that the health system is capable of adopting new modes of detection, appropriately and in ways which enhance, rather than undermine, equity. The considerations identified by the roundtable group point to a wide range of areas for further research, analysis and careful policy deliberation. However, the group also identified three main priorities for action now:

Updating the taxonomy of diagnosis

The scope of possible diagnoses is constantly evolving to reflect new techniques for detecting diseases and ways of describing them more precisely. But are we asking one word – diagnosis – to do too much? Diagnosis is often used to describe the process of identifying disease no matter its current manifestation or likely progression. It is important to distinguish this from screening which, by contrast, seeks to identify disease, or increased risk, in apparently healthy individuals. At the same time, we are increasingly aware of the uncertainties inherent in diagnosis and screening – including limitations in predicting disease or clinical outcomes for a given individual – but communicating these continues to be a challenge. There is arguably a need to reconsider how we describe and categorise physiological phenomena that occupy a space close to the boundary between the normal and the pathological.

Key questions to explore:

- Do we need new terminology to describe the diverse activities currently captured under diagnosis, and the different uncertainties associated with these?
- Do we need to establish clearer boundaries between screening, early detection and diagnosis?

Strengthening evaluations of the impact of diagnostic tests on clinical outcomes

Concerns were raised around the evaluation of the use of tests and if there was sufficient evidence informing implementation. Systems exist for the evaluation of tests in different contexts (e.g. NSC for screening, ACCE for genetic testing and the NICE Diagnostic Advisory Committee). Current health data infrastructures do not allow for routine collection and continuous evaluation of diagnostic tests, locally or nationally. This limits scrutiny of the use of diagnostic tests particularly related to patient outcomes.

Key questions to explore:

- What evidence is needed to determine that early diagnosis informed by biological insights is positively impacting patient outcomes?
- What evidence do we need to decide if a diagnostic test should be implemented, and what outcomes matter?
- Given this, the question becomes how to ensure system-wide buy in to enable, in an ideal scenario, ongoing evaluation of diagnostic tests?

Communicating diagnosis in context

Diagnosis requires effective communication, both between healthcare professionals and with patients. The diagnostic process increasingly requires multi-disciplinary working, in which different clinicians need to be able to communicate the contributions and limitations of their specialist knowledge to the construction of a diagnosis. Even specialities associated with a higher degree of 'diagnostic objectivity', such as radiology, require a certain amount of interpretation. A test result is not in itself a diagnosis and so the role of the clinician is to put these results in the context of the individual patient. Communication needs to support knowledge exchange with patients and equip them with understanding to promote shared decision-making and autonomy.

Key questions to explore:

- How can clinicians most effectively communicate the limits of diagnostic tests?
- How can public engagement be used to understand a wider range of perspectives and preferences for diagnosis and the diagnostic process?

5. What next?

Diagnosis is a broad term that has different meanings depending on the clinical context and for patients. Technological advances offer opportunities for early diagnosis for many diseases, and the future of healthcare seeks to harness this innovation for patients. In a resource constrained health system, adoption of these technologies requires increasingly sophisticated and multidisciplinary clinical pathways.

How diagnosis intersects with emerging technologies, the use of data, and transformation of health services is of ongoing significance. This roundtable was an important first step to identify key challenges and areas for action arising from the changing nature of diagnosis. This change is likely to accelerate in the future, and we and others need to be alert to this. Going forward, it will be crucial to involve other perspectives – including patient and public views – to take this work forwards.