The changing nature of diagnosis

Background briefing

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Advances in medical imaging and molecular testing, including genomics, as well as in data-driven analysis and artificial intelligence, introduce novel opportunities for investigating the causes of ill health and detecting biological signs of disease before the onset of symptoms. The consequences of these developments for the diagnostic process, including its scope, timing, precision and degree of certainty, warrants focussed multidisciplinary attention.

To begin to address this, the PHG Foundation, working with Dr Zoë Fritz, has convened this roundtable discussion to generate and capture views on how diagnosis is changing in response to technological innovation. This discussion is intended as an initial step in a process aimed towards unpicking how increased interaction between technology and medical diagnosis impacts on patients, healthcare professionals and the health system.

We aim to understand how different forms of information (driven by diverse sources) contribute to the construction of a diagnosis, particularly in contexts where a significant level of information is probabilistic and inherently uncertain. Our immediate objective is to probe the stability of diagnosis in light of biomedical innovation with a view to identifying priorities for future research, policy and practice in this area.

Scope

We are interested in the role of diagnosis in different medical contexts and across a wide range of specialities. However, we do **not** intend to discuss psychiatric diagnoses or medically unexplained symptoms in this roundtable. This is not to ignore the interconnectedness of physical and psychological symptoms but to recognise that these topics introduce added complexity.

This background briefing introduces some of the topics and concepts that we will discuss in the roundtable.

What is diagnosis?

Diagnosis can be thought of as both:

- **a classification system:** 'a pre-existing set of categories agreed upon by the medical profession to designate a specific condition;'[1] and
- **a process:** 'a complex, patient centered, collaborative activity that involves information gathering and clinical reasoning with the goal of determining a patient's health problem.'[2]

1 Jutel, Annemarie. 'Sociology of Diagnosis: A Preliminary Review'. *Sociology of Health & Illness* 31, no. 2 (2009): 278–99. https://doi.org/10.1111/j.1467-9566.2008.01152.x.

2 Balogh, Erin P., Bryan T. Miller, and John R. Ball, eds. *Improving Diagnosis in Health Care*. Washington, D.C.: National Academies Press, 2015, p. 32. https://doi.org/10.17226/21794.

Conventionally, the diagnostic process begins when an individual with symptoms seeks medical attention. This initiates a period of information gathering, synthesis and interpretation, which may draw upon a combination of methods including clinical history and interview, physical examination, diagnostic testing and consultation with specialists. An initial list of potential causes - a differential diagnosis - may be updated and refined as further information is acquired, until a definitive diagnosis can be reached.

Diagnosis is related to, but distinct from, other processes in the clinical pathway such as early detection and screening. However, as screening tests become more common, the boundaries between the two may become blurred and challenge the conventional model of symptomatic patients seeking medical attention. For example, cholesterol levels can be used to diagnose hypercholesterolemia and as a tool to screen for risk of cardiovascular disease.

Questions to consider:

- *Are there other ways of considering diagnosis, or aspects which are particularly important in your field?*
- *How should boundaries between detection and early diagnosis be understood?*

What is the purpose of diagnosis?

Diagnosis aims to provide an accurate understanding of a patient's health condition and in doing so guide appropriate clinical decision making and improve opportunity for a positive outcome.[3] It simultaneously acts as a shorthand that supports effective communication and referral within the health system. This guides treatment and management decisions by diverting patients into defined care pathways. In epidemiology, diagnosis can help to understand the distribution and determinants of disease in populations, enabling the identification of risk factors and patterns of disease occurrence. For patients, diagnoses are important for meaning-making, to understand their situation, explain the situation to others, plan for the future and to get access to treatment and care.

The purpose of diagnosis may differ depending on how the concept of disease is viewed. Two classically opposed framings are:

- Nominalism: Under this approach, diseases are defined by the symptoms, signs, and patterns observed in patients, and the naming of diseases is a way to categorise and communicate these observations. Diagnosis, therefore, is a form of labelling - a shorthand used by physicians to classify 'abnormal' biology so that they can better manage their patients and determine their prognosis with greater accuracy.
- Essentialism: The essentialist view argues that each 'disease' has an underlying fixed essence or form. It is a real entity, pre-categorised within the body. Under this framing, diagnosis is seen as the act of discovering 'disease' in the body.

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3 Balogh, Erin P., Bryan T. Miller, and John R. Ball, eds. *Improving Diagnosis in Health Care*. Washington, D.C.: National Academies Press, 2015, p. 31. https://doi.org/10.17226/21794.

It is important to consider that social norms play a role in determining what counts as a diagnosis and subsequently what the impact of this is on the patient. These vary over time and across cultures and, alongside advances in biomedical knowledge and technology, they are a relevant factor when evaluating what to diagnose and when.

The changing nature of diagnosis

Over the last 50 years, there has been a vast increase in the number of diagnostic categories. International Classification of Diseases (ICD) codes[4] have rapidly expanded to include more than 50,000 in the most recent version (ICD-11).[5] This increase in the number of diagnostic categories allows for more granular differentiation between disease subtypes but introduces more complexity in making a diagnosis as more specific conditions are recognised.

Questions to consider:

- *What is driving this trend and what are its consequences?*
- *In which contexts might it be more or less useful to have more precise, granular categories?*

Increasingly sophisticated technological and data-driven innovations offer the opportunity to better identify and diagnose patients. These have become integral to clinical pathways and pivotal to ongoing medical decision making. Examples include[6]:

- *In vitro* diagnostics: Genomics, biosensors, radiomic tests, and molecular diagnostics
- Imaging/radiology: MRI, X-ray, PET and CT scans
- Digital products, apps and software: clinical decision support, predictive algorithms, software as a medical device (notably AI-powered diagnostic and clinical support tools), and wearables

The diagnostic process is evolving in response to technological innovation. These changes may arise in different ways, depending on the context. Data generating technologies allow clinicians to more quickly and accurately answer a wide range of clinically impactful questions, for example, through employing rapid metagenomics to accurately inform diagnosis of sepsis.[7] This time-sensitive context must consider feasibility if value is to be achieved. Additionally, innovation (for example in AI technologies) could provide an opportunity to restructure traditional diagnostic pathways. Importantly, each technology

4 ICD is a system of categorisation and coding for disease

5 Hofmann, Bjørn. 'Too Much, Too Mild, Too Early: Diagnosing the Excessive Expansion of Diagnoses'. *International Journal of General Medicine* 15 (6 August 2022): 6441–50. https://doi.org/10.2147/IJGM.S368541.

6 Powell, D., Hannah, A. The dichotomy of diagnostics: exploring the value for consumers, clinicians and care pathways. npj Digit. Med. 7, 101 (2024). https://doi.org/10.1038/s41746-024-01087-8

7 Charalampous T, Alcolea-Medina A, et al. Routine Metagenomics Service for ICU Patients with Respiratory Infection. Am J Respir Crit Care Med. 2024 Jan 15;209(2):164-174. https://doi.org/10.1164/rccm.202305-0901OC

comes with limitations and clinical judgement is needed to interpret test results within the context of a patient's individual history, symptoms and overall health picture. For example, under a proposed new model for stroke diagnosis and treatment referral, an AI tool produces a recommendation, based on processing MRI/CT imaging, which then must be assessed alongside clinical judgment and conventional imaging.[8]

Questions to consider:

- *How are advances in technologies, including the way in which they have been developed and implemented, impacting on diagnostic practice?*
- *In which context(s) is earlier diagnosis of disease likely to be most beneficial? Does this come at the cost of accuracy and certainty?*

How is this impacting different clinical contexts?

The suitability of advanced diagnostics is dependent on the clinical context and the disease in question. Some diagnoses present with a higher degree of certainty, while most diseases are considered as a continuum. Clinical experts define thresholds and, where criteria are met, a patient is considered to have disease. There is a trade-off between diagnostic accuracy and timeliness and the balance of these choices is determined by the clinical context. Different clinical contexts may fulfil different roles in the diagnostic process:

- *Primary care:* In this context, precise diagnostic labels are often less important than determining the correct course of action. Diagnoses may be framed in terms of dichotomous decisions: treatment versus non-treatment, referral versus non-referral, and serious versus non-serious.[9] Increasingly, the focus of clinical time is being redirected towards risk, not symptoms, which raises questions around whether, and in what contexts, it is appropriate to 'diagnose' risk.[10]
- *Acute and emergency medicine:* Patients present with conditions often characterised by rapid onset, severe symptoms, of life-threatening nature. Emergency rooms (ERs) assign a preliminary diagnosis to patients, which may change when they are hospitalised. Patients' needs are increasingly complex, particularly from growing multimorbidity and polypharmacy.
- *Outpatient services and specialist care:* As diagnostic tests become more targeted and refined, specialists must navigate increasingly complex diagnostic pathways, often involving multimodal data integration (for example, vital signs, imaging and lab tests, such as genomics) and requiring collaboration across specialties. It may also lead to a reconfiguration of clinical roles, triggered, by example, through advances in AI technology.

Questions to consider:

- How does the role of diagnosis differ across clinical contexts?
- How does the clinical context impact the requirements for data generating technologies?

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8 D'Adderio, L., Bates, D.W. Transforming diagnosis through artificial intelligence. npj Digit. Med. 8, 54 (2025). https://doi.org/10.1038/s41746-025-01460-1

9 Summerton N. Making a diagnosis in primary care: symptoms and context. Br J Gen Pract. 2004 Aug;54(505):570-1.

10 Martin, Stephen A., et al. 'Sacrificing Patient Care for Prevention: Distortion of the Role of General Practice'. BMJ 388 (21 January 2025): e080811. https://doi.org/10.1136/bmj-2024-080811

Challenges in diagnosis

Emerging technologies are positioned to change diagnosis. Our background research has highlighted the following concerns, but we are keen to understand your perspectives on these and other challenges.

Diagnosing too much, too mild, or too early

Diagnosis has many important functions and holds significant benefit for individuals, professionals, and for societies. However, excessive diagnosing could be harmful. Hofmann describes this in terms of three categories: too much, too mild and too early.[11]

- **Too much** diagnosis can occur when diagnostic labels are applied to phenomena not previously considered diagnostic, e.g. mildly elevated blood pressure or obesity, in a way that may not benefit the individual and can sometimes cause harm.
- Lowering diagnostic thresholds to include milder cases in the definition of the disease or in its diagnostic criteria (i.e. being **too mild**), can lead people to be diagnosed with diseases that may not have caused symptoms or harm. Gestational diabetes and early-stage chronic kidney disease may serve as examples.[12] While detecting and treating milder cases may appear successful, many of the subsequent treatments may be unnecessary.
- Diagnosing **too early** may identify abnormalities that never progress to disease, leading to overtreatment and anxiety (e.g. ductal carcinoma in situ, indolent lesions of epithelial origin, lung nodules on a CT scan).

Diagnostic uncertainty

Diagnostic uncertainty has been described as the "subjective perception of an inability to provide an accurate explanation of the patient's health problem."[13] The clinician's intention is to build enough confidence in a hypothesis to optimise clinician decision-making. Uncertainty emerges in the context of diagnosis in both 'knowable' and 'unknowable' ways.[14] An example of the former is the probability of a given diagnostic test being accurate. The latter refers to the imperfect nature of the current state of knowledge.

Uncertainty in diagnosis can be exacerbated by many factors. Of particular relevance, diagnosing at an earlier stage can increase uncertainty, for example when abnormal biology is detected before the development of symptoms and it cannot be known whether this will progress into symptomatic disease. Managing and communicating diagnostic uncertainty in this changing landscape is challenging, both logistically and ethically.

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11 Hofmann, Bjørn. 'Too Much, Too Mild, Too Early: Diagnosing the Excessive Expansion of Diagnoses'. *International Journal of General Medicine* 15 (6 August 2022): 6441–50. https://doi.org/10.2147/IJGM.S368541.

12 Moynihan R, Brodersen J, Heath I, et al. Reforming disease definitions: a new primary care led, people-centred approach. *BMJ Evid Based Med*. 2019;24(5):170–173. doi:10.1136/bmjebm-2018-111148

13 Bhise, Viraj, Suja S. Rajan, Dean F. Sittig, Robert O. Morgan, Pooja Chaudhary, and Hardeep Singh. 'Defining and Measuring Diagnostic Uncertainty in Medicine: A Systematic Review'. *Journal of General Internal Medicine* 33, no. 1 (January 2018): 103–15. https://doi.org/10.1007/s11606-017-4164-1.

14 Krishnan, Lakshmi, Yvonne Commodore-Mensah, Kelly T. Gleason, David P. W. Rastall, David E. Newman-Toker, and Kathy McDonald. 'Roads Diverge: Mapping the Journey towards Diagnostic Health Equity'. *BMJ Open Quality* 14, no. 2 (20 April 2025). https://doi.org/10.1136/bmjoq-2024-003135.

Various strategies for managing diagnostic uncertainty have been reported, including communication methods such as 'safety-netting'[15], ordering additional investigations, monitoring to allow signs and symptoms to develop, and consulting external sources of information such as risk calculation tools.[16][17]

Question to consider:

- How can clinicians and patients best be supported in communicating and understanding diagnostic uncertainty?

Inequity

Several characteristics of diagnosis could potentially lead to inequity.

- Geographic and socioeconomic disparities mean that some individuals - especially in low-income, rural, or underserved areas - have limited access to diagnostic tools, specialists, or even primary care. While differential access to treatment is more transparent and readily measurable once a condition is identified, diagnostic disparities often remain invisible within healthcare systems.[18]
- Marginalised groups are more likely to experience underdiagnosis, late diagnosis, or misdiagnosis due to systemic barriers and biases. For example, AI algorithms have been found to underdiagnose conditions in underserved populations, such as Hispanic female patients, due to training on non-representative datasets.[19] This is a problem, as diagnosis acts as a gateway to accessing care.
- When a diagnosis is correct, the implications (e.g. access to treatment, stigma, insurance impact) can still vary widely depending on a person's socioeconomic status, immigration status, or ability to advocate for themselves within the healthcare system.
- While treatment decisions can be framed as choices to enable shared decision-making, the diagnostic process leaves little room for co-production, especially where patients have lower levels of education, literacy, or experience language barriers. Additionally, the amount of information shared by clinicians, and its degree of certainty, may be influenced by assumptions about a patient's educational background and tolerance for risk.

Question to consider:

- How might changes in diagnostic practices impact different population groups? In which contexts might it exacerbate or mitigate against inequities?

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15 This refers to advice given to patients to help them to identify when they need to seek further medical attention.

16 Cox, Caitríona L, Benjamin M Miller, Isla Kuhn, and Zoë Fritz. 'Diagnostic Uncertainty in Primary Care: What Is Known about Its Communication, and What Are the Associated Ethical Issues?' *Family Practice* 38, no. 5 (1 October 2021): 654–68. https://doi.org/10.1093/fampra/cmab023.

17 Fritz, Zoë, and Richard Holton. 'Too Much Medicine: Not Enough Trust?' *Journal of Medical Ethics* 45, no. 1 (1 January 2019): 31–35. https://doi.org/10.1136/medethics-2018-104866.

18 Lakshmi Krishnan, Yvonne Commodore-Mensah, Kelly T Gleason, David P W Rastall, David E Newman-Toker, Kathy McDonald - Roads diverge: mapping the journey towards diagnostic health equity: BMJ Open Quality 2025;14:e003135.

19 Seyyed-Kalantari L, Zhang H, McDermott MBA, Chen IY, Ghassemi M. Underdiagnosis bias of artificial intelligence algorithms applied to chest radiographs in under-served patient populations. Nat Med. 2021 Dec;27(12):2176-2182. doi: 10.1038/s41591-021-01595-0.

What is the future of diagnosis?

This briefing has provided a short introduction into what is meant by 'diagnosis', how the practice of diagnosis is changing in response to technological development and some of the challenges raised.

In this roundtable, we will also explore the future of diagnosis. We are interested in whether diagnostic practices, and the terminology surrounding diagnosis, are fit for purpose in this increasingly complex system where we have more advanced testing modalities, earlier detection and a shift in patient expectations, and what might need to change in light of this.

Question to consider:

- What do you see as the biggest challenge facing the practice of diagnosis? What are the key priorities in addressing this?