Reforming disease definitions: a new primary care led, people-centred approach

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Abstract

Expanding disease definitions are causing more and more previously healthy people to be labelled as diseased, contributing to the problem of overdiagnosis and related overtreatment. Often the specialist guideline panels which expand definitions have close tis to industry and do not investigate the harms of defining more people as sick. Responding to growing calls to address these problems, an international group of leading researchers and clinicians is proposing a new way to set diagnostic thresholds and mark the boundaries of condition definitions, to try to tackle a key driver of overdiagnosis and overtreatment. The group proposes new evidence-informed principles, with new process and new people constituting new multi-disciplinary panels, free from financial conflicts of interest.

Introduction

Expanding definitions of disease are causing too many people to be diagnosed and treated unnecessarily, producing harm and waste, posing a major threat to human health and the sustainability of health systems, and creating growing conflict within medicine.[1 2] For example, the widely used definition of 'chronic kidney disease' labels around half of all older people, yet many of them will never experience related symptoms.[3] Changes to the definition of gestational diabetes could double its prevalence, despite a lack of clear evidence that the expansion will bring the newly diagnosed meaningful benefits that outweigh harms.[4] Recently, a new definition of hypertension which labels one in every two adults, while welcomed by some, has been soundly rejected by family doctors over concerns it may cause more harm than good to many people.[5] Responding to growing calls for action to address this key driver of overdiagnosis, from professional societies and other groups around the globe,[2 6 7] we are proposing a new primary care-led, multidisciplinary, independent, people-centred approach to defining disease.

The unmet need for reform

An ongoing series about overdiagnosis in the *BMJ* has documented global concern about expanding disease definitions and the subsequent risk that many people may be unnecessarily diagnosed and overtreated, across numerous and diverse conditions, including attention deficit hyperactivity disorder,[8] osteoporosis[9] and pulmonary embolism.[10] Evidence suggests the specialist guideline groups which regularly review disease definitions often decide to expand them: by lowering thresholds to capture more people at lower risk of future illness; by creating pre-diseases; by overmedicalising common or mild life experiences; or by changing diagnostic processes.[11] Moreover, while perhaps driven by the best of intentions, many existing guideline groups do not rigorously investigate the potential harms of their proposals to expand definitions,[11] reflecting the wider problem of under-investigation of harms in medicine.[12 13] These specialist-led guideline groups also often include many individuals with financial ties to pharmaceutical companies, some of which benefit directly from expanded definitions.[11] The guideline which launched the broad new condition called 'chronic kidney disease' was sponsored by Amgen, and despite sustained criticism about the potential for overdiagnosis, the broadened definition has been defended and reaffirmed by individuals and groups with ties to pharmaceutical companies.[14]

Changes to disease definitions driven by disease-specialists understandably reflect their desire to detect and intervene early—and not miss a needed diagnosis—often by overmedicalising people at lower risk, hoping to prevent onset of the serious illness they see daily in specialist practice. By contrast, family or generalist doctors—who are currently under-represented on panels which change disease definitions—witness daily how specialist-driven definitions turn too many healthy people into patients, contributing to the overload of primary care systems with un-resourced demand.[15] As a result, general practice groups are at the forefront of calls for solutions. The Royal College of General Practitioners has a standing group on overdiagnosis, the Royal Australian College of General Practitioners has joined a call to address overdiagnosis,[16] while the Nordic Federation of General Practice has lead advocacy on the issue within the World Organisation of Family Doctors (WONCA), whose member

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Box 1 New principles to underpin development of disease definitions

New processes
- Informed by new Guidelines International Network's guidance and checklist.
- Explicit sensitivity to potential harms of overdiagnosis.
- People-centred.
- Independent of commercial interests.

New people
- Generalist, primary care led—informed by disease-specialists.
- Citizen representation.
- Multidisciplinary.
- Independent of commercial interests.

organisations represent around half a million family doctors globally.

WONCA Europe's landmark 2018 declaration states 'overdiagnosis means making people into patients unnecessarily…by medicalising ordinary life experiences through expanding definitions of disease' while noting 'underdiagnosis and overdiagnosis may exist side by side'.[2] WONCA's call to action arises from its definition of general practice: 'a key role for the discipline is to provide advocacy, protecting patients from the harm which may ensue through unnecessary screening, testing, and treatment…'.[17] As the Danish College of General Practitioners recently articulated, key aspects of the family doctor role are to treat the sick, let the healthy stay healthy, work for timely diagnoses and avoid overdiagnosis (J Brodersen, personal communication, 2018). Our aspiration is to see diagnoses offered to those who will benefit from them, rather than those for whom they may cause more harm than good.

New principles, new processes, new people

Responding to a growing mood for reform, explicit calls to end specialist dominance of disease definitions,[6] and work already underway, we are proposing a new set of principles, involving new process and new people (see box 1).

New processes

In 2017, members of the Guidelines International Network's (G-I-N) Overdiagnosis Working Group—initiated by generalist family doctors—proposed a new process for those seeking to modify disease definitions, and it published a world-first evidence-informed guidance.[18] In developing the guidance authors noted they had been 'unable to identify any currently accepted criteria for modifying a disease definition', highlighting both the variation in how definitions are changed and how there are no rules governing such critically important processes. The new checklist outlines eight domains to examine explicitly before proposing changes to definitions, including the number of people affected, potential benefits for the newly diagnosed, potential harms and the balance between benefits and harms. Using the checklist to examine the 2017 expanded definition of hypertension, researchers concluded that while some people at high risk would benefit, a majority of the newly diagnosed could only experience harm from an unnecessary label and potential overtreatment.[19] In addition to working with this new guidance, we believe the process of disease definition needs much greater explicit sensitivity to the potential harm of overdiagnosis. Against a backdrop of the rise of a new 'person-centred' approach,[20] any assessments of potential benefits should focus, wherever possible, on meaningful outcomes that matter to people, rather than surrogate markers. Finally, to maximise trust, decision-making processes should listen to, but be free from, individuals or organisations tied financially to industries with interests in the outcomes.

New people

Along with new processes, there is a clear need for new people to constitute panels which recommend diagnostic thresholds and mark the boundaries of condition definitions. Existing G-I-N standards suggest panels should be multidisciplinary and 'include diverse stakeholders, such as health professionals; content experts; methodologists with skills in evidence appraisal and synthesis; and, ideally, healthcare consumers and health economists'.[21] With their generalist approach, specialists in primary care may be uniquely placed to lead new multidisciplinary teams to define disease, including disease-specific specialists, citizen representatives, members from social sciences, and others who can help connect the process more to the civil societies ultimately impacted. All members would be independent of financial ties to industry—as has been recommended by high level reports[22]—or professional financial self-interest. Like other models for independent decision-making,[23] it is vital that non-conflicted and broadly representative panels seek evidence and testimony from more narrow specialist interests, which may include those with professional or financial conflicts.

Reforming disease definitions and diagnoses

Informing the reform with evidence

Important to this reform will be evidence of the extent of overdiagnosis arising from inappropriately expanded disease definitions. There is a need for estimates of how many people are currently being diagnosed unnecessarily—across common conditions—accompanied by estimates of the consequent burden of harm and waste. While there are ongoing debates about optimal methods, research quantifying the extent of overdiagnosis across cancers is well established, but new research is needed to quantify estimates across non-cancer conditions. Work is already underway on optimal methods for estimating overdiagnosis for non-cancer conditions,[24] and this work will inform projects within individual conditions, which may ideally be undertaken within a global collaboration. Informed by this evidence, our proposed new approach will have both broad and more focused implications.

Delayed diagnosis and de-diagnosis

In a broad sense, this proposal is designed to create more debate and scientific investigation of diagnoses, their benefits and harms, and how they might best work for people within the structure of health systems.[25] As well as finding new processes and new people to set diagnostic thresholds, it may be timely to re-imagine these as *thresholds for discussion* with potential patients.[26] This may be particularly relevant where there is controversy and uncertainty around different diagnostic thresholds, and different people will have different perspectives on the appropriateness of a label or treatment—for example, with the condition currently described as mild hypertension, where evidence suggests treatment of people at low risk may do more harm than good.[27] A related reform, proposed for some psychiatric diagnosis,[8] is the idea of more routinely delaying a diagnosis until it is clearly necessary. Similarly, it is

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time to investigate ways to make diagnoses more temporary, where appropriate, and less fixed in stone, and to explore methods for de-diagnosis, in the same way researchers and clinicians have developed methods for de-prescribing.[28 29] Just as the practise of the 'Medication Review' can help people reduce unneeded medications, a new form of 'Diagnosis Review', carried out by a family doctor for those regarded as having multiple morbidities may in some cases enable reduction of unneeded labels and treatments. The aim is to ensure diagnoses are in place only when there is a degree of certainty they will bring more benefit than harm.

Partner organisations for reform

In a more focused sense, our aim is to initiate new processes with new people, who will ultimately produce newly reformed definitions of individual diseases, conditions, diagnostic criteria and thresholds. Organisations which may auspice or promote such reform include WONCA, the preventing overdiagnosis group, G-I-N and/or national primary care organisations, potentially working in alliance with other professional, civil society or public agencies such as the WHO, a co-sponsor of the 2018 preventing overdiagnosis conference. Sometimes a reformed definition may simply propose raising a diagnostic threshold to de-medicalise those at low risk or with mild problems. On other occasions, a rigorous review of evidence may warrant an entirely new approach, as has been proposed for the risk factor for fracture currently described as a disease called osteoporosis, a construct arbitrarily created by a group supported by pharmaceutical companies.[9]

De-medicalising risk

The medicalisation of being 'at risk' of a future disease axiomatically creates overdiagnosis, as some people at risk will never suffer that disease, but instead only experience long-term harms of an unnecessary label and treatment. The lower the threshold at which risk is medicalised, the higher the numbers of people diagnosed unnecessarily. Many of the entities that have become known as 'diseases' or 'chronic conditions'—including high blood pressure, high cholesterol, diabetes type 2 and osteoporosis—are more correctly understood as states of being at risk. Bizarrely, the increasing creation of 'pre-diseases' in some cases medicalises those who are at risk of being at-risk, for example, pre-diabetes.[30] Our proposed reform will investigate ways to de-medicalise the risks of the healthy, while maintaining an appropriate emphasis on public health and prevention, requiring courageous, careful and lateral thinking, broadly and condition-by-condition.

Conclusion

Developing a framework for this long-term reform and facilitating a global collaboration to enact it will involve proactive and reactive efforts that we hope will drive a cultural shift and a practical change in how diseases are defined. Research teams will continue to quantify estimates of overdiagnosis arising from current disease definitions, informing priorities for action. Actions include the constitution of new panels, with new processes and new people, to review and revise existing definitions. Concurrently, primary care organisations will become more reactive to expansions in definitions seen as increasing the risk of overdiagnosis, such as the controversial 2017 hypertension widening, explicitly rejected by the American Academy of Family Physicians, and other groups,[5] and the rejection of the expanded definition of gestational diabetes by the Royal Australian College of General Practitioners.[31] An international meeting to review progress on our proposal and develop more detailed strategies for change will take place at the December 2019 *Preventing Overdiagnosis* conference in Sydney

There are important limitations, uncertainties and caveats to note as we propose this ambitious reform of disease definitions, which will provoke opposition from those whose markets are directly threatened. First, we write as a group working across a multitude of influential national and international organisations, but we do not in this instance represent them. Second, our backgrounds and thinking are largely medical, and there is clearly opportunity for this initiative to be informed by evidence, experience and theories outside medicine, including, for example, from philosophy.[20] Third, addressing the problem of expanding disease definitions is but one of many potential solutions to overdiagnosis,[7] and much important work is underway already to try and wind back the harms of too much medicine, safely and fairly, such as calls to action within our associations,[2] creation of new medical curricula, scientific discussion at national and international meetings and new information materials for the public. Fourth, given the novel nature of this proposal, there is not yet a mature evidence-base to support it. Fifth, there is clear synergy between this proposal and the calls for reform of clinical practice guidelines,[32] which has not been explored in this analysis. And finally, we acknowledge moves to expand definitions, to detect and treat people earlier, are often driven by the best of intentions, and we see great merit in identifying those who will benefit from a medical label and subsequent care. However, notwithstanding the good intentions driving a bad system, the human person can no longer be treated as an ever-expanding marketplace of diseases, benefitting professional and commercial interests while bringing great harm to those unnecessarily diagnosed.

Key messages

- Expanding definitions of disease are a key driver of overdiagnosis and related overtreatment, while specialist panels proposing these expansions are often conflicted and do not investigate potential harms.
- A leading international primary care-led group is responding to growing calls for action to address this problem, and is proposing a new way to define disease, as one way to reduce overdiagnosis.
- New processes will involve using explicit guidance to assess potential benefits and harms when modifying disease definitions, with a focus on people-centred outcomes, and new panels may be primary-care led, multidisciplinary, with representation from civil society and independent from financial ties to industry.
- Next steps include research quantifying the extent of overdiagnosis across key conditions, and developing and evaluating this new approach to defining disease in different settings.

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Correction notice This article has been updated since its original publication to correct the spelling of author name of Minna Johansson.

Contributors The authorship team comprises 12 general practitioners and one non-medical health researcher. The general practitioners include both practising and academic family doctors, from countries across three continents, Europe, Australia and Latin America, representing a range of different perspectives. The authors include general practitioners who work at senior levels, across a range of influential national and international organisations, including the World Organization of Family Doctors, but do not in this instance represent them. All authors have a strong interest in the problem of expanding disease definitions, a key driver of overdiagnosis. The paper arose from discussions within this group, as well as a small meeting held in Copenhagen in August 2018, during the Preventing Overdiagnosis international scientific conference, based on the evidence and analysis cited. The first author, RM, is guarantor.

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