RACGP
Royal Australian College of General Practitioners

Expansion of disease definitions

When the definition of a disease expands as the result of emerging clinical evidence, it can result in:

- changes to the thresholds and criteria for a diagnosis of the disease
- an increase in the number of people with a diagnosis of the disease,[1–4] which can cause an apparent increase in its prevalence.

Although changes to a definition are sometimes required to ensure patients receive the treatment they need (eg for a fast-developing condition that requires early diagnosis), they can sometimes lead to low-value care or harm to patients, their families and the health system.

Why the expansion of a disease's definition can be a concern

The following table lists factors that can contribute to an expansion of a disease's definition, and the possible concerns relating to each factor.

Factor	Possible concerns
Lack of consistent standards to define what is a 'disease'	Although guideline developers generally refer to this checklist when considering expanding a disease definition, currently there are: • no international standards for the membership of panels or committees that define diseases • inconsistent protocols for handling conflicts of interest • no agreed criteria for reviewing disease definitions.[4]
Expert and clinical guideline groups	Although expert and clinical guideline groups generally change disease definitions and thresholds with good intentions, changes can result in unintended consequences – for example, if members have financial or academic conflicts of interest that unintentionally influence the group's decisions.[1,3]
Risk factors being classified as a disease	Although it is common for risk factors for chronic conditions to be treated as if the risk factor itself is a disease,[1] treatments that improve risk factors but don't actually improve a patient's health are often low value. An example is prescribing antihypertensives for a person with mildly raised blood pressure but otherwise low overall risk of cardiovascular disease.

Factor	Possible concerns
Genetic risk factors being classified as a disease	Genetic risk factors, which typically only affect a subset of people, should not be labelled as a disease. Examples include the DQ7/DQ8 genetic marker for coeliac, or *ApoE4* gene for Alzheimer's disease, neither of which guarantee a person will develop the disease.
'Medicalisation of life'	Labelling physiological and/or emotional changes that occur naturally to most people as a 'disease' is often of no benefit.[1] Examples include: • the fall in testosterone levels in men being labelled the andropause[1] • uncomplicated recent grief being diagnosed as a mental health disorder.[5]
Increased sensitivity of screening and testing	Although technological advances have resulted in more sensitive tests that may pick up 'milder' versions of a disease, labelling or treating this mild/early disease may cause more harm than good[4,6] – for example, sensitive one-stop blood glucose criteria resulting in a diagnosis of gestational diabetes. For further information on screening and testing sensitivity, refer to Overdiagnosis.
The belief that diagnosing a disease early always leads to better outcomes	Although early diagnosis and treatment is beneficial for patients with some diseases, it can sometimes lead to unnecessary harms, particularly if the patient does not benefit from the treatment.[7] Furthermore, evidence does not support claims that early diagnosis helps patients improve their lifestyle.[3,8]
Increased labelling of people in the early stages of a disease, or at high risk of a disease	There is no evidence that labelling people with a 'pre' disease (eg prediabetes, pre-dementia, pre-hypertension) benefits the patient.[9]

Harms from expanding disease definitions

Harms to the patient, their families, and the health system as a result of changing disease definitions include the following.

- **Harms resulting from overdiagnosis**
  Refer to Overdiagnosis.
- **Physical, psychological and financial harms to patients and their families**[2,6]
  Patients may experience the following harms, particularly if their symptoms are mild and/or they are at low risk of future illness:[10]
  - physical – side effects from unnecessary medications and procedures[4,6]
  - psychological – the patient's belief that they are suffering from a disease can increase their anxiety about their health, affect their identity, and result in stigma from others[2,6,10]
  - financial – these include out-of-pocket costs for treatment, time off work, increased health insurance costs, ineligibility for health insurance.

Family members can also experience anxiety and distress worrying about the patient and themselves (eg worrying that they may also have the disease), may need take time off work to support or care for the patient, and can be affected by the additional costs.

- **Potential lack of benefit of treatments for the expanded group of patients**
  Treatments that were designed and tested for patients with the original definition of the disease may be administered to patients who now meet the criteria for the disease's expanded definition.[9] However, these treatments may result in little or no benefit for these patients and may cause more harm than benefit.
- **Increased demands on the healthcare system**
  When people with a milder disease are diagnosed with a more serious disease, this places increased demands on the healthcare

system, which means fewer resources for those with a more urgent need[7] and an increase in waiting lists, particularly for people in rural and regional areas.

- **Harms to the environment**
  Increasing the number of people receiving unnecessary treatments increases the environmental impact of healthcare.[11] Although there is a lack of data about the current carbon footprint of Australian general practice,[11] estimates from the United Kingdom suggest that between 65% and 90% of the general practice carbon footprint is the result of the prescribing of pharmaceuticals,[11,12] and 9% the result of pathology testing and diagnostic imaging.[11,13]

How you can manage expanding disease definitions

Apply a critical lens to the expanded definition.

When reviewing an expanded disease definition, ask yourself:[9]

- How does the expanded disease definition differ from the previous one?
- Why do we need an expanded definition?
- How will it change the number of people diagnosed?
- What are the potential benefits for patients diagnosed according to the expanded definition?
- What are the potential harms for patients diagnosed according to the expanded definition?
- How useful is the expanded definition at predicting clinically important outcomes?
- How precise and measurable is the expanded definition?

Consider the outcomes of diagnosing a patient with this disease.

- How will this diagnosis help them?
- How could this diagnosis harm them?
- How might this diagnosis affect their family/carers?
- Will this diagnosis change how the patient is managed?

Undertake shared decision-making with your patient.[2]

Discuss with your patient and/or their carer what a diagnosis label means for them. Consider including the following in your discussion:

- how the definition of the disease has changed
- that they might not have been diagnosed with the disease before the change to the definition
- the physical, psychological and financial consequences of receiving a diagnostic label
- the potential harms and benefits of
- treatment, which may include medication or invasive procedures
- using evidence-based non-drug interventions
- watchful waiting

Consider evidence-based non-drug interventions where indicated.

Consider using evidence-based non-drug interventions and progressing to more intensive or invasive treatment only if the non-drug interventions are ineffective. This approach may be particularly helpful for people who fall into a 'pre' category, or might not have been diagnosed with this disease before the change to the definition.

For further information on non-drug interventions, refer to the RACGP's *Handbook of non-drug interventions (HANDI)*.

Consider watchful waiting where appropriate.[6]

Discuss watchful waiting with your patient to decide if it is preferable, given their circumstances. Your discussion should include the potential benefits and harms of treating a disease they might not have been diagnosed with before the change to the definition.

Further reading

RACGP position statement

- Too much medicine

RACGP *First Do No Harm* GP resources

- Overdiagnosis
- Overtesting

Other resources

- Sustainable healthcare | Cochrane
- Guidance for modifying the definition of diseases: A checklist | Doust J, Vandvik PO, Qaseem A, et al. *JAMA*.
- 1 in 6 women are diagnosed with gestational diabetes. But this diagnosis may not benefit them or their babies | Glasziou P, Doust J. *The Conversation*
- We need new rules for defining who is sick. Step 1: Remove vested interests | Moynihan R, Glasziou P. The Conversation
- How to rein in the widening disease definitions that label more healthy people as sick | Moynihan R, Horvath R. The Conversation

Information for patients

- 5 questions to ask your doctor or other healthcare provider before you get any test, treatment, or procedure | Choosing Wisely Australia
- Questions to ask a health professional | Wiser Healthcare
- What is overdiagnosis? | Wiser Healthcare

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ID-6139