VIEWPOINT

LESS IS MORE

The Impact of Definitions of Disease on Overdiagnosis

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The concept and definition of disease influences both clinical practice and public health. The World Health Organization has defined health but failed to define disease. The contemporary concept of disease emerges from biology but is influenced by social, cultural, and economic factors. Definitions are important, as how we label a condition can determine how society perceives, manages, and supports patients. Continuously expanding disease criteria can reduce underdiagnosis and increase appropriate care, but often risk overdiagnosis, resulting in overtreatment and low-value care, ultimately threatening health care sustainability.

There exists significant disagreement about what constitutes a disease.[1] Differences in how diseases are conceptualized influence changes in disease criteria. From a naturalist point of view, diseases are objective and harmful deviations from the normal functioning of the body. In contrast, a constructivist perspective emphasizes social norms and economic interests driving our conceptualizations. Chronic fatigue syndrome, fibromyalgia and post–COVID-19 condition are examples of diseases that are difficult to delineate because their biological underpinnings remain uncertain.

In the presence of such uncertainty, disease criteria may evolve over time, periodically giving rise to new conditions. New diseases may arise, for example, from new causes (COVID-19), advances in diagnostic technologies (early-stage cancers), or conscious advocacy (chronic fatigue syndrome).[2] On the other hand, criteria for existing diseases may evolve, as has been the case with diabetes since the 1960s.[3] Psychiatry has seen many such changes, as each revision of the *Diagnostic and Statistical Manual of Mental Disorders* has added conditions and changed criteria for established conditions. Guideline panels defining disease thresholds often broaden definitions, thus expanding the population labeled as having a particular disease. To ensure guidelines are evidence-based, practical, and sustainable, panels should include not only experts from narrow subspecialties but also generalists, primary care physicians, and other health care professionals who likely better understand real-world constraints, as well as patient partners.

Each person defines their health individually, and this perception may differ from a physician's assessment. Some perceive themselves as healthy despite severe pathology, while others perceive themselves as ill even in the absence of identifiable abnormalities. A population-based survey revealed considerable differences in perceptions of what constitutes a disease among groups of laypersons, physicians, nurses, and parliament members.[1] Of 60 conditions surveyed, only 12 (20%), including diabetes and pneumonia, were considered diseases by at least 80% of respondents. In contrast, 5 (8%), including aging and homosexuality, were considered not to be diseases by at least 80% of respondents. For most conditions, including erectile dysfunction, drug addiction, and menopause, large disagreements existed. Physicians were more likely than laypersons to label conditions as diseases, likely reflecting their focus on pathophysiological mechanisms.

Table. Impact of Definitions (Cutoff Points) on Disease Prevalence

Analyte	Cutoff	Prevalence, %
Total cholesterol, mg/dL	180	54
	200	35
	220	20
	240	10
Fasting glucose, mg/dL	100	61
	110	30
	120	15
	126	12
Systolic blood pressure, mm Hg	110	75
	120	50
	130	27
	140	14

SI conversion factors: To convert cholesterol to mmol/L, multiply by 0.0259; glucose to mmol/L, multiply by 0.0555.

Prevalences calculated from National Health and Nutrition Examination Survey 2017-2020 data of adults 20 years or older more in the US. Examination weights were used for total cholesterol and blood pressure. Fasting weights were used for fasting glucose and low-density lipoprotein cholesterol. For systolic blood pressure, the mean of three consecutive measurements was used. Low-density lipoprotein cholesterol was estimated using the Friedewald equation.

Although attention to patient perspectives is key to good clinical care, reliance on individual perspectives to define diseases may prove insufficient. Secondary gains, such as financial compensation, along with advertising campaigns that inflate the perceived severity of common conditions (such as dry eye[4] or overactive bladder syndromes[5]), can shape perceptions of illness. Social stigma may further contribute by reinforcing these perceptions.

Driven by commercialism, direct-to-consumer advertising, and the pursuit of perfect health—a culture of "more is better"—modern medicine has fostered a culture of overdiagnosis and overtreatment. To avoid this harmful approach, clinicians should remain alert to commercial and social influences and practice shared decision-making, balancing patient values with evidence-based disease thresholds. Resources like *JAMA Internal Medicine*'s Less is More series,[6] the international Choosing Wisely campaign,[7] and *The BMJ*'s Too Much Medicine series[8] can provide valuable guidance.

Medicalization, the framing of nonmedical issues in medical terms, easily leads to overuse of tests and treatments with little benefit but significant harm and cost. Overdiagnosis, related to medicalization, labels conditions that would never have caused symp-

toms or shortened life as diseases. Cancer screening programs often detect slow-growing tumors that would never have caused harm, leading to resource-intensive active surveillance programs and unnecessary treatments. As a result, the impact of screening on overall mortality for many types of cancer, including breast and prostate cancer,[9] remains uncertain. Increased sensitivity of imaging may increase the detection of incidental abnormalities in joints, disks, and blood vessels that might be better left uninvestigated. Advances in artificial intelligence and genetic testing may also contribute to overdiagnosis.

Changes in diagnostic thresholds for conditions such as hypertension, diabetes, and osteoporosis reflect a tendency to expand disease definitions, increasing the population labeled as diseased. For example, wider criteria for gestational diabetes have doubled its prevalence without improving maternal or neonatal health outcomes.[10] While there is a legitimate role for measuring and treating risk factors such as hyperlipidemia, hypertension, and hyperglycemia, lowering diagnostic thresholds can lead to overdiagnosis, overtreatment, and a drastic increase in prevalence in health care systems already struggling with sustainability (**Table**). In psychiatry, broadened criteria risk pathologizing normal behaviors, such as redefining shyness as social anxiety disorder or everyday restlessness as attention-deficit/hyperactivity disorder. These broadened definitions may result in problematic medication use and harmful labeling.

While recognizing more conditions as diseases can improve access to treatments, it also raises the possibility that people with manageable variations of normal life may come to view themselves as sick. Disease labels can worsen professional prospects, particularly where diagnoses affect insurance or employment. Expending resources on essentially healthy individuals also leads to opportunity costs, including delayed care for those most in need. This issue is global, as expanded screening, modern imaging, and skewed financial incentives contribute to overdiagnosis and overtreatment in both high-income and low- to middle-income countries. To combat these issues, researchers must prioritize studies evaluating whether broader disease definitions translate into better patient outcomes or simply more prescriptions and procedures.

The evolving definition of disease shapes clinical care and public health, often medicalizing normal life variations and contributing to overdiagnosis. Physicians must discern whether symptoms stem from treatable conditions or are part of normal life, in the process ensuring diagnoses improve health outcomes and quality of life. The global challenge of defining disease underscores the need to balance broader access to medical treatment with avoidance of harmful medicalization and inefficient resource use.

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