



Expanding disease and undermining the ethos of medicine

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Abstract

The expansion of the concept of disease poses problems for epidemiology. Certainly, new diseases are discovered and more people are treated earlier and better. However, the historically unprecedented expansion is criticised for going too far. Overdiagnosis, overtreatment, and medicalization are some of the challenges heatedly debated in medicine, media, and in health policy making. How are we to analyse and handle the vast expansion of disease? Where can we draw the line between warranted and unwarranted expansion? To address this issue, which has wide implications for epidemiology, we need to understand how disease is expanded. This article identifies six ways that our conception of disease is expanded: by increased knowledge (epistemic), making more phenomena count as disease (ontological), doing more (pragmatic), defining more (conceptual), and by encompassing the bad (ethic) and the ugly (aesthetic). Expanding the subject matter of medicine extends its realm and power, but also its responsibility. It makes medicine accountable for ever more of human potential dis-eases. At the same time it blurs the borders and undermines the demarcation of medicine. Six specific advices can guide our action clarifying the subject matter of medicine in general and epidemiology in particular. To avoid unlimited responsibility and to keep medicine on par with its end, we need to direct the expansion of disease to what effectively identifies or reduces human suffering. Otherwise we will deplete medicine and undermine the greatest asset in health care: trust.

Keywords Concept of disease · Expansion · Diagnostics · Overdiagnosis · Philosophy · Responsibility · Ethics · Epistemology · Ontology · Aesthetics

Introduction

With the great advances in science and technology disease is expanded in a number of ways. From the 2400 diseases classified in Sauvage's *Nosologia methodica* of 1768 [1] to the 45,000 disease codes the WHO's International Classification of Disease of ICD-10 [2] there is a huge extension of medicine. There was a corresponding increase in the number of disorder categories in DSM from 182 in 1968 to 297 in 1989 [3], in ICPC's categories [4], as in ICF [5]. Hence there has been a substantial expansion of disease categories and entities with the effect that more people than ever are diagnosed with a disease [6].

Part of this expansion¹ is due to a historically unprecedented extended knowledge of diseases, their mechanisms, and their causes. We know more about what causes pain and suffering than ever before. At the same time we have gained insights in risk factors, processes, precursors, and indicators of disease. Although only some of these may cause pain and suffering, we identify and handle them as disease.

No doubt, the expansion has many positive consequences, for example in differentiating existing diseases in more precise and actionable disease entities—or such as diagnosing, treating, and helping more people—earlier. However, the expansion also has some negative implications such as overdiagnosis, overtreatment, medicalization, as well as adverse effect of futile diagnostics and treatment and increased anxiety and altered self-conception from becoming diagnosed. Some attention has been given to the side-effects of expanding disease definitions, and guidelines to ensure appropriate expansions have been suggested [7, 8]. How these can address the unprecedented number of new biomarkers, risk factors, and indicators that emerge from the convergence of

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¹ With expansion I mean increasing the extension of disease.

omics, Big Data, Artificial Intelligence and enormous investments [9] is still to be decided.

Expanding disease

The expansions of disease come in six fundamentally different types: by expanding or knowledge of disease mechanisms (epistemic); by extending the phenomena that define disease (ontologically); by what we can do (pragmatic); by what we define as disease (conceptual); by what we define as bad (ethic); and by what we consider to be ugly (aesthetic). Figure 1 illustrates the dimensions of disease expansion.

Table 1 gives an overview of various ways that disease is expanded with its implications.

Expanding disease by expanding knowledge (epistemic)

Disease is expanded through the great increase in medical knowledge. On the positive side, increased knowledge increases the numbers of diseases by establishing new disease entities or by dividing existing diseases in other and more precise disease entities. However, extended knowledge also has some potentially less favourable effects on the extension of disease. First, more knowledge of mechanisms, processes, precursors, and risk factors of

disease may make us look harder and find more cases, and extend the number of persons being diagnosed with a disease. Second, the same can happen when introducing more precise tests [10]. New methods for detecting pulmonary embolism (high-resolution CT scan) greatly enlarged the extension of pulmonary embolism [11–13]. Third, increased attention or precision may make us lowering diagnostic detection thresholds [14–16], further increasing the number of persons being considered to have the disease. This connects to the fourth way that knowledge expands disease: increased precision makes us find milder cases. When more milder cases are detected and treated, success rates increase [17], spurring more emphasis on testing and further expanding disease. The epistemic extension relates to an ontological expansion of disease.

More things are made disease (ontological)

Disease is expanded by making more things count as disease. This can happen in several ways. First, new phenomena can come to define diseases directly. This can be phenomena such as behaviour (ADHD), emotions (sorrow, dysphorias), sensations (dry eye), or hormones (Low-T). While finding new things that can explain and define disease is crucial in medicine, e.g., in the discovery of microbes, making new things disease is also at the core of the type of expansion that is called medicalization. Second, new phenomena can define disease indirectly, through various indicators, such as biomarkers (beta-amyloid), risk factors (blood pressure), and precursors (polyps, HPV). These phenomena may come to be included in the definition of the disease, as in the case with beta-amyloid and Alzheimer's disease. This expansion can be both helpful and harmful.

Doing more: defining more (pragmatic)

“Can do–will do” seems to be another strong drive in expanding disease. When a condition can be manipulated or treated, it rapidly is made a disease. Bariatric surgery has contributed making obesity a disease. Being able to measure and manipulate blood pressure and cholesterol levels has made these conditions classified as diseases (hypertension and hypercholesterolemia). As already mentioned, the effects of lowering treatment thresholds reinforce this effect. No doubt, the ability to do more can certainly be good, but not always, e.g., in cases where conditions unnecessarily are identified as diseases or where it implies more harms than benefits.

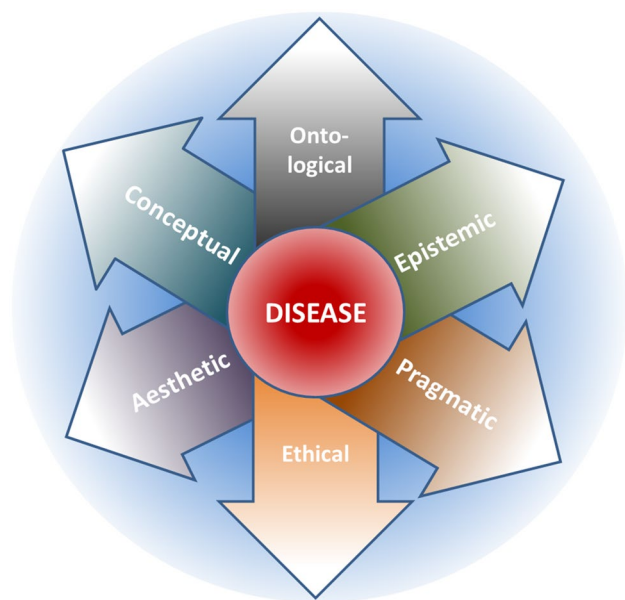


Fig. 1 Six ways of disease expansion

Table 1 Various ways of expanding disease with potential positive and negative implications and examples

| Type of expansion | Way of expansion | Example | Potential positive implication(s) | Potential negative implication(s) |
|---|---|---|---|---|
| Knowing more about what can be made disease (epistemic) | Increased knowledge of mechanisms, processes, precursors and risk factors | Polyps, infectious agents | Avoiding disease (primary prevention) | Adverse side effects from treatment, overtreatment |
| | More precise tests | Beta-amyloid, CT-detected pulmonary emboli, High-sensitivity cardiac troponin | Gaining knowledge of causes, treatment | |
| | Lowering diagnostic thresholds (cut-offs) | Diabetes Blood pressure Cholesterol level | More accurate diagnosis | Overdiagnosis |
| Making more things disease (ontological) | New (experienced or observed) phenomena define disease | Male baldness, menopause, sexual function, sorrow, low-T, dry eye, drapetomania, ADHD | Reducing underdiagnosis | Overdiagnosis, indication creep, subsequent overtreatment |
| | Indirectly defining disease by indicators, such as biomarkers, precursors, and risk factors | Biomarkers, such as troponin, glucose control, atypical ductal hyperplasia, osteopenia, microcalcifications | Increased help | Medicalization |
| Pragmatic | Increased actionability: can do, will do | Obesity, hypertension, hypercholesterolemia | Avoiding consequences of disease (secondary prevention) | Overdiagnosis, overtreatment |
| Conceptual | Expanding theories and models of disease (including indolent and regressive cases) | Ductal carcinoma in situ (DCIS), cervical intraepithelial neoplasia | Preventing disease and suffering | Side effects, harm |
| Ethical expansion | Being directed by our moral imperative to help | Deviant behavior | Preventing disease and suffering (secondary prevention) | Overdiagnosis, overtreatment |
| Aesthetic | Including deformities (which do not hamper function) | Pectus excavatum, protruding ears, bags under eyes | Increased help to persons and groups of persons | Medicalization Anxiety Stigmatization |
| | | | Aesthetic normalization | Reinforcing aesthetic norms and values, side effects of treatment |

Expanding definitions (conceptual)

Expanding theories and models of disease (including indolent and regressive cases) also make us expand disease. For example, extended knowledge about breast cancer developments made ductal carcinoma in situ (DCIS) defined and treated as a disease (cancer). While the conceptual expansion is significant, new initiatives urge us to reconsider [18]. For example it has been suggested to rename DCIS indolent lesions of epithelial origin (IDLE) [19]. Other conceptual changes can be found with papilloma and grade 1 carcinoma of the bladder, cervical intraepithelial neoplasia, and non-invasive encapsulated follicular variant of papillary thyroid carcinoma [20]. How we define (and how we name) conditions also expands disease. This can be for the good (if the extended definition includes what is harmful) and for the bad (if it includes conditions or states that are not related to human suffering).

The bad and the mad (ethic)

Yet another way that disease is expanded is by our moral imperative to help people. Making various types of behaviour, such as criminality and gambling, disease in order to take it out of the social and into the medical realm was thought to protect or help people. Despite potentially good moral intentions, the results are controversial. Other cases, such as drapetomania and dissidence are examples of where what was considered to be morally wrong behaviour was made disease [21].

The ugly and the ill (aesthetic)

Correspondingly, aesthetics has contributed in expanding disease. Aesthetic norms have formed professional norms. What is ugly has become disease. Protruding ears and funnel breast are treated on the level with any other disease. Moreover, cleft lips and palates are treated with good aesthetic results. Despite good intentions of helping people, the making of something as a disease has also lead to stigmatization and discrimination. Hence, the common and often well founded (aesthetic, but also ethic) impetus to help persons with specific characteristic or conditions, drive us to make them diseased.

The complex expansion

Clearly, these ways of expanding disease are neither exclusive nor exhaustive. There are obvious overlaps. For example obesity has become a disease because we can treat it

(pragmatically), because we want to help people (ethic) [22] and due to our norms of beauty (aesthetics). Correspondingly, there are overlaps in implications of expanding disease as well. Overtreatment may result from overdiagnosis (epistemic or conceptual expansion) or from making new phenomena disease (ontological expansion). Moreover, medicalization can occur due to both ontological expansion and due to ethic expansion. There are certainly also other ways of expanding disease than those mentioned here, e.g., by making certain ideologies disease. This article focuses only on the main types in order to be able to identify and contain unwarranted expansion.

Expanding disease and extending responsibility

No doubt, modern medicine appears as a tremendous success, addressing a wide range of crucial human challenges. Thus, medicine appears as an attractive problem-solver for ever more human problems. This provides ample opportunities to the various forms of expansions discussed above. Clearly, expanding disease gives medicine increased influence and amplified power, but expanding its realm also extends its responsibility and liability [23, 24].

Hence, when disease becomes limitless, so does medicine's accountability. Becoming responsible for ever more human phenomena and types of unease may become a bigger bite than medicine can chew. Moreover, accountability presupposes demarcation. You can only be "liable to be called to account" for matters that are reasonably delimited. This calls for concern with the reduced delimiting function of disease.

Inflating disease: depleting medicine and its major asset

By unwarranted expansion of disease medicine broadens and blurs its borders. When "anything and everything" becomes disease, the concept of disease cannot be used to demarcate medicine [25].

In this there is a risk of decoupling disease from the suffering of people [26]. Ever more conditions called disease are not experienced by persons as (bodily or mental) suffering. Hence, implicit in the expansion of disease there is a transgression of the traditional end of medicine, i.e., to help suffering people. This may well be seen as a modernizing and positive development of medicine, pre-empting suffering, but it can also be a crucial change in its ethical foundation. As eloquently pointed out by Iona Heath:

Box 1 Specific advice

- | | |
|----|--|
| 1. | Only knowledge that establishes a clear relationship to human suffering should be allowed to expand disease |
| 2. | Only phenomena that can be clearly related to human suffering should be included in definitions of disease |
| 3. | Only those potential actions that effectively identify or reduce human suffering should be allowed to define disease |
| 4. | Only change definitions of disease where it can be documented to effectively identify or reduce suffering |
| 5. | Do not let the need to help people dictate the expansion of disease as other ways to help may be more effective |
| 6. | Do not confuse ugly with diseased. Avoid treating social (aesthetical) norms with medical means. Avoid making aesthetical norms govern professional values |

we have, for the first time in history, separated our notions of disease from the human experience of suffering and have created an epidemic of disease without symptoms, defined only by aberrant biometrics. An ever greater proportion of healthcare resources are directed towards reducing these numbers to some fictitious state of normality and, in the process, those who are perfectly well are not only assigned labels, that in themselves can be shown to compromise health, but are also exposed to treatments with significant adverse effects. [27] (my emphasis)

However, nobody can be responsible for everything. What is at stake is the core resource of medicine: trust [28, 29]. When expanding disease beyond where we clearly can demonstrate to help people, we may undermine the trust in medicine. When we cannot inform people about whether they will suffer from what is identified as disease, or their true chances of being saved or overdiagnosed and overtreated, people may lose their trust in medicine altogether. That is, when doubting whether disease labels will help or harm us, we will stop relying on the labels and the health professionals administering them.

Harnessing medicine through its core concept, disease

There are of course other ways that medicine enlarges its extension than expanding disease. Engaging in social determinants of health is but one example. Enhancing immunity with vaccines is yet another [30] and gene editing a third. In this article the focus has been limited to the expansion of disease. Correspondingly, a wide range of drivers of this expansion have been identified in the literature [30–34] and are crucial in avoiding uncontained expansion.

Moreover, the overzealous expansion of medicine has fuelled a series of *counter-movements*, such as Choosing Wisely, Too Much Medicine, Less is More, Overuse, Smarter Medicine, Preventing Overdiagnosis, Slow Medicine and other campaigns (<http://www.lessismoremedicine.com/projects/>). Other measures have been used to delimit medicine

from “non-medical issues.” For example, social prescribing has been implemented to shift demand for non-medical needs from primary care to communities [35] and deprescribing has gained increased attention. Despite such laudable efforts, their effectiveness in delimiting medicine still appear modest. For example, the efforts to disinvest and decommission low-value care have been moderate or low [35–40].

One reason for this may be that these efforts do not target what is considered to be the ethos of medicine, such as the concept of disease related to human suffering. Therefore, to (de)limit medicine by (de)limiting disease can be a fruitful strategy as it ties medicine to its ultimate end. Before de-diagnosing disease, it appears to be crucial to contain the ontological, epistemic, pragmatic, conceptual, aesthetic, and ethical expansion of disease in order to avoid depleting medicine. Box 1 summarises advice specific to each type of expansion.

However, reaffirming that (identifying and reducing) *suffering* is the key goal of medicine, and a key (moral) notion of demarcation for expanding disease does not mean that all people’s experienced sufferings are imperative to medicine. Suffering is only a necessary condition, it is not sufficient. The point is that suggestions to expand disease in any of the six dimensions that have been addressed here can be dismissed if the expansion does not relate to human suffering in any identified and verified way.

This does not mean that we have to exclude cases of identified conditions which we cannot yet treat. Finding a new infectious agent may certainly warrant the expansion of disease even if we cannot (yet) treat the condition. The key point is whether it is related to suffering in an effective and verified way. It has to identify or reduce human suffering in some demonstrable manner.

Conclusion

Disease is expanding in several ways: by increased knowledge (epistemic), making more phenomena count as disease (ontological), doing more (pragmatic), *defining more (conceptual)*, and by including the bad (ethic) and the

ugly (aesthetic). Expanding the subject matter of medicine extends its realm and power, but also its responsibility. It makes medicine accountable for ever more of human potential dis-eases. At the same time it blurs the borders and undermines the demarcation of medicine. In expanding disease medicine may become limitless and transgressing its traditional ultimate goal, i.e., to help suffering people. To avoid unlimited responsibility and to keep medicine on par with its end, we need to restrict expansion of disease beyond where it identifies or reduces human suffering. Otherwise we will deplete medicine and undermine the greatest asset in health care: trust.

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