

The concepts of ‘health’ and ‘disease’

Underlying assumptions in the idea of value in medical interventions

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Abstract

This chapter provides an overview of the problem of conceptual definition of ‘health’ and ‘disease’ as a background to situate and introduce the discussion about health outcomes and value of new medical interventions. This work reflects and discusses broader literature on this topic by highlighting the available – and lacking – definitions in the specific context of health institutions in the UK. After introducing the World Health Organization’s (WHO) definition of ‘health’ and some general criticisms of it, two more recent and particularly relevant positive definitions of ‘health’ are presented, stressing the potential connection between such definitions and the role of healthcare services. This is followed by a more extensive analysis of positive definitions of ‘disease’ since the technical literature seems particularly prolific and relevant. In order to organise the discussion, this chapter frames the different approaches to positive definitions of ‘disease’ within the fact/value problem. At the same time that it introduces three of the most influential conceptualisations of disease (Biostatistical Theory, The APA Task Force work and the Harm Dysfunction Analysis), it illustrates three possible positions regarding the fact/value problem in this matter (strong descriptivism, strong normativism, and mixed descriptive/normativism, respectively). Finally, because of the lack of a successful and agreed definition of ‘disease’, this chapter highlights recent efforts to embrace the disjunctive and vague elements of this concept, allowing and encouraging specific and contextual cluster definitions of ‘disease’, which seem particularly useful to contextualise and open the discussion on how to think about the idea of health outcomes and value in medical interventions.

Keywords: philosophy of medicine, concept of ‘health’, concept of ‘disease’, line-drawing problem, contextual definitions, goals of medicine

1 Introduction: building the bridge between the concepts of 'health' and 'disease' and the of idea value in medical interventions

Conceptual constructs tend to become naturalised and their meanings are taken for granted in the work to push disciplines further. Although this might be necessary to some extent, keeping in mind the frailty of conceptual definitions may be just as important for disciplines to move forward without oversimplifying or lacking context and complexity. Thus, by taking a step back and focusing on the question of what is value in the context of new medical interventions, this book highlights the importance of conceptual discussions in the background of highly applied fields associated with healthcare delivery which face challenging practical decisions of prioritisation, resource allocation and conflicting goals.

Although this is an issue of rich discussion – further explored in other chapters of this book – 'value' in healthcare is understood in general terms as patient health outcomes achieved per money spent, and so, it is argued to encompass many healthcare goals (Porter, 2010). However, the idea of health outcomes itself is a huge topic of discussion and this way of operationalising health, although needed, also faces significant challenges, not just on how to define them, but also how to measure them. In the specific context of the United Kingdom (UK), the National Health Service (NHS) has developed an outcomes framework to measure institutional progress. The framework is organised around five key dimensions:

- preventing people from dying prematurely
- enhancing the quality of life for people with long-term conditions
- helping people to recover from episodes of ill health or following injury
- ensuring that people have a positive experience of care
- treating and caring for people in a safe environment and protecting them from avoidable harm (NHS, 2020).

Although analysing these dimensions in-depth goes beyond the scope of this work, acknowledging this framework serves the purpose of showing how sanitary goals and health outcomes can potentially raise tensions when prioritising.

It is clearly unfeasible to pursue all these outcomes simultaneously and some may come into direct conflict with one another. A good example of this is how sometimes enhancing the quality of life of someone living with a long-term condition may imply that it is not possible to prevent that person from dying prematurely.¹

¹ Parsons' contribution in this book discusses this precisely. The author argues that all patients (including those who lack decision-making capacity) ought to sometimes forego dialysis in favour of conservative kidney management, prioritising their quality of life over life-extending treatment.

Complexities associated with this are in direct connection with their immediate conceptual context, i.e. the concepts of 'health' and 'disease' to which this chapter is dedicated. Academic discussion around these two underlying concepts in all health-related issues is highly prolific and still unresolved. Usually taken for granted, one could claim these concepts may be central to defining when, how and with which goals medical interventions should be developed and applied, framing and contributing to the overarching focus of this book: exploring different definitions and approaches to value and how to incorporate these into the assessment of new medical interventions.

This chapter will argue that there needs to be a clear rationale connecting the concepts of 'health' (and 'disease'), health outcomes and the value of medical interventions to have consistent systems with clear and achievable goals in which results or measurements can actually be put into context and offer valuable input. We will see how this core idea underlies most of the problems presented in this book.²

The concepts of 'health' and 'disease' play an important part in everyone's daily experience of being alive and still manage to escape the descriptive possibilities of language – puzzling philosophers of science, sociologists, psychologists and many others, these concepts somehow invite Augustine's reflection on the question of "what, then, is time?" – so sensibly highlighted by Ricoeur in the preface to *Time and Narrative*, "I know well enough what it is, provided that nobody asks me; but if I am asked what it is and I try to explain, I am baffled" (Saint Augustin in Ricoeur, 1984, p. xi). Very similarly, addressing the concepts of 'health' and 'disease' implies engaging in an ongoing quest.

² Chapters in this book, which refer to this conceptual consistency issue, are those by Buch *et al.* on highly-priced pharmaceuticals and by Steigenberger *et al.* on integrating patients' and social aspects into Health Technology Assessments. In the first case, assessing whether certain pharmaceuticals are too expensive will depend heavily on what societies are willing to pay and this, in turn, might be argued to depend greatly on what the states of health and disease are. This latter point also applies to the second case, which focuses on patients' perceived value of the quality and benefit of a health technology. More contributions in this book focused on economics, such as those by Himmler and Mitchell, exemplify and refer to this matter. These works use specific understandings and frameworks for the monetary value of health in terms of well-being, for example, those defined by a specific measurement of quality of life (quality-adjusted life years) (Himmler, in this volume). Mitchell's work (in this volume) challenges this notion, as the quality of life captured by quality-adjusted life years might be considered too narrow, suggesting a shift from outcomes focused on health (such as quality-adjusted life years) towards people's capabilities.

2 The complexities behind conceptualising health: Its operationalisation and the goals of medicine

The current Constitution of the NHS in the UK, last updated in 2015, does not define what counts as health or disease. However, it does state that the NHS' aims to improve health and well-being, supporting people to keep mentally and physically well, to help them get better when they are ill and, if they cannot fully recover, help them to stay as well as possible till the end of their lives (NHS, 2015). It is possible to see from this statement that the core idea underlying the use of the concept of 'health' mirrors a positive definition³ based on a state of well-being that includes both a mental and physical dimension. The UK's National Institute for Health and Care Excellence (NICE) does not define health specifically but health-related quality of life as "a combination of a person's physical, mental and social well-being; not merely the absence of disease" (NICE, 2018). The NICE definition paraphrases one stated in and enforced by the WHO Constitution, according to which, "health is a state of complete physical, mental and social wellbeing and not merely the absence of disease or infirmity" (WHO, 1948). In this manner, the UK seems to embrace a positive understanding of health, rejecting negative definitions based on the absence of disease.

The *overall* assessment of the definition offered by the WHO proposes that by suggesting a positive operationalisation of the concept of 'health', it represents an improvement over previous negative definitions, but it, nonetheless, raises significant problems. These are mostly related to the idea of "complete well-being", which NICE omits.

Critical views on the definition offered by the WHO raise issues particularly relevant for the topic of this book. Some critiques suggest that it seems idealistic and unachievable, labelling most of the population as unhealthy most of the time. Critiques argue that this definition could contribute to the medicalisation of society, justify unlimited development of drugs or treatments, and create serious challenges for healthcare systems that have to find a balance between individual health needs and the resources available. Further critiques of this definition are related to disease patterns shifting from acute to chronic conditions, supporting the idea that conceptualisations of health and disease might be closely related to historical context and associated health developments. Additional critiques of this definition include, among many others, problems for disease classifications systems (e.g. quality of life, disability, functioning)

³ In general terms, this means a definition focused on what health is instead of what it is not. Health defined as the absence of disease, for example, is usually described as a negative definition of health.

since health, as “complete” well-being, does not allow for measurement or operational specification (Bircher, 2005; Huber *et al.*, 2011).

In all critiques, these considerations work to bridge the gap between the relevant conceptualisations of health and disease and define the value of medical interventions. Broadly speaking, if the value of medical interventions comes from a focus on improving the health and reducing the disease burden on individuals and populations, then what counts as ‘health’ and ‘disease’ matters.

Although there is still no consensus on a satisfactory positive definition of the concept of ‘health’, or even on whether this is possible or desirable (Boorse, 2011), many interesting new definitions have arisen from the discussion. I will introduce two rather recent positive definitions of health that serve the purpose of illustrating a broader and more nuanced view on this issue, stimulating insightful reflection for this particular work. Bircher (2005) suggests conceptualising health as,

a dynamic state of wellbeing characterized by a physical, mental and social potential, which satisfies the demands of a life commensurate with age, culture, and personal responsibility. If the potential is insufficient to satisfy these demands the state is disease. (Bircher, 2005, p. 336)

This definition seems to be an overall improvement of the definition offered by the WHO because it allows health to be a variable state within the lifespan of an individual, attending to relevant dimensions and being, in this sense, more realistic. However, this definition resorts to controversial or difficult concepts – mental and social potential and personal responsibility – that would also require a definition for this concept of ‘health’ to be practicable.

In contrast to this long-winded definition, Huber *et al.* (2011, p. 2) define health as the “ability to adapt and self-manage”, with specific characterisations in the three domains of health: physical, mental and social. This understanding of health seems particularly interesting because it diverges completely from the WHO legacy, stressing the capacity or functioning of the individual, thus, potentially reconfiguring the role of healthcare services and the value of medical interventions in terms of support towards developing such functioning.

Furthermore, this definition may be particularly relevant in current times when chronic diseases are the main disease burden in the UK and the rest of the world. Chronic diseases currently account for 90 % of all deaths in the UK, and the risk of dying prematurely from a chronic disease is 11 % (Office for National Statistics, 2017; WHO, 2017).⁴ Furthermore, chronic diseases account for 50 % of all general practice appointments, 64 % of outpatient appointments, 70 % of all inpatient bed days and 70 % of the total health and care expenditure

⁴ These statistics are based on the four main groups of non-communicable chronic diseases (cardiovascular disease, cancer, diabetes and obstructive pulmonary disease).

in England (Department of Health, 2012).⁵ This information implies that currently and in terms of disease burden, full recovery is not an option in most cases, therefore, self-management and the possibility to mobilise resources become key concepts in assessing the value of medical interventions (Bodenheimer *et al.*, 2002). This idea of self-management and the possibility to mobilise resources remains closely linked to the capabilities approach and its application to this field.⁶

3 The complexities behind conceptualising disease: what can we learn from key definitions?

Regarding positive definitions of ‘disease’, those that do not merely place explanatory value on the absence of health, the overall picture is just as dynamic and unresolved. This discussion is very prolific both in terms of the literature generated and the many working definitions (Lemoine, 2013; Walker and Rogers, 2018). However, specifically in the context of official health institutions in the UK, neither the NHS nor NICE acknowledges or defines of the concept of ‘disease’ or any other related concepts such as ‘disorder’, ‘condition’, ‘sickness’, ‘infirmity’ or ‘illness’. Therefore, it might be thought that such national institutions implicitly embrace a negative definition of ‘disease’ by setting their focus on health. In other words, ‘disease’ is broadly taken to be the absence of health.

As an effort to systematise the extensive literature on the concept of ‘disease’, Boorse (2011) suggests that there are five commonly present elements in most ‘health’ and ‘disease’ definitions. These elements include (1) medical treatment, (2) pain, discomfort and disability, (3) statistical abnormality, (4) disvalue and (5) specific biological ideas: homeostasis, fitness and adaptation. However, counter-examples for each of these elements show that all fail to be neither necessary nor sufficient for a satisfactory definition of these concepts at an abstract theoretical level,⁷ thus, illustrating how challenging it seems to be to reach satisfactory definitions.

⁵ These statistics are based on a category of chronic diseases that is not restricted to non-communicable diseases, though it includes the main four groups as well (cardiovascular disease, cancer, diabetes and obstructive pulmonary disease).

⁶ This is discussed in-depth later in this book in the chapters by Mitchell – briefly mentioned previously – and Ubels, who highlights the importance of combining information about capability, functioning and utility in the assessment of the value of medical interventions.

⁷ Counter-examples for each element include (1) all disease for which there is no treatment available and, conversely, non-disease medical treatments, such as plastic surgery or contraceptive pills; (2) pathological conditions that do not involve pain, discomfort or disability, such as hypertension, and, conversely, non-pathological conditions that may involve all or some of these elements, such as pregnancy; (3) many statistically abnormal

Traditional conceptual analysis in philosophy broadly implies aiming at an exact, descriptive definition by “testing a definitional criteria and exceptions against a set of given cases, while drawing up counter-cases against an opponent’s definition”, thus, identifying conditions that are both necessary and sufficient to define a concept and the exceptions to these conditions (Lemoine, 2013, p. 310). However, because of the lack of a satisfactory descriptive definition of ‘health’ and ‘disease’, it has been argued that conceptual analysis can provide *descriptive* or *naturalist* (factual) definitions or *normativist* (value) definitions. The former are value-free definitional criteria, while the latter are value-laden definitional criteria, broadly assuming that disease is bad for the person and health is desirable. Although most authors provide some kind of normativist definitional criteria for ‘health’ and ‘disease’, which may be soft, in the sense that may also include some descriptive conditions, some very influential definitions adopt a strong descriptive approach, stressing the importance to continue working on value-free definitional criteria for ‘health’ and ‘disease’ (Boorse, 2011; Lemoine, 2013).

In order to illustrate this very dynamic discussion and provide some background on what is the state-of-the-art, it seems relevant to present some of the most influential definitions of the concept of ‘disease’. These include Boorse’s (1977) *Biostatistical Theory*, which represents a strong descriptivist (value-free) position, Spitzer and Endicott’s (1978) tentative proposed definition and criteria of medical and mental disorder, which represents a rather normativist (value-laden) position, and Wakefield’s (1992) *Harm Dysfunction Analysis*, which represents a mixed position, stressing the importance of naturalist and normativist components.

Boorse’s (1977) *Biostatistical Theory*, being largely laden towards normativist definitions of ‘health’ and ‘disease’, emerges as a strong critique of previous literature on the topic. The author offers a strong descriptive definition, stressing that health and disease evaluations are sensitive to contextual and indivi-

conditions are not diseases, such as being left-handed and, conversely, many statistically normal conditions involve a pathological condition such as gum disease or tooth decay; and (4) depending on the context, a disease may not necessarily be bad for the individual. An example of this would be that flat feet during a period of war might save someone’s life by precluding them from joining the armed forces, thus, potentially being regarded as a good thing. Finally, (5) many non-disease human functions are not homeostatic, such as growth or reproduction, and, conversely, pathologies such as sterility do not produce any homeostatic failure. If one considers that fitness stands for individual survival and reproduction, many pathological conditions do not interfere with these, such as anosmia, and, conversely, many non-pathological activities, such as mountaineering, may increase the risk of early death. Regarding adaptation, depending on the context, some diseases may not be maladaptive. An example of this would be a severe immune deficiency in a sterile environment (plastic bubble) and, conversely, many non-pathological conditions may be adaptive in one context and not in another, such as being light-skinned in Iceland or in Africa (Boorse, 2011).

dual variables, highlighting that the conceptual definition should be value-free to allow the individual to value the condition according to relevant specific circumstances. In this manner, according to the *Biostatistical Theory*, ‘health’ is defined by normal functioning, where what is normal is statistically determined and functioning refers to biological functions. Furthermore, ‘disease’ consists of deviations from the species’ biological design, therefore, identifying ‘disease’ is considered a matter of natural sciences rather than an evaluative judgment. Thus, the overall rationale and assumptions underlying this definition imply four main criteria: (1) definition of the reference class (an age group of a sex of a species), (2) definition of normal function within members (based on a statistically typical contribution to the individual survival and reproduction), (3) definition of ‘health’ in a member of the reference class as a normal functional ability and (4) definition of ‘disease’ as an internal state which reduces functional abilities below typical efficiency (Boorse, 1977).

Spitzer and Endicott (1978), building on their previous work as members of the American Psychiatric Association Task Force on Nomenclature and Statistics (which suggested a largely criticised first definition of the concept of medical and mental disorder in 1976), provide a revised definition of these concepts, which states that,

a medical disorder is a relatively distinct condition resulting from an organismic dysfunction, which in its fully developed or extreme form is directly and intrinsically associated with distress, disability, or certain other types of disadvantage. The disadvantage may be of a physical, perceptual, sexual, or interpersonal nature. Implicitly there is a call for action on the part of the person who has the condition, the medical or its allied professions, and society. A mental disorder is a medical disorder whose manifestations are primarily signs or symptoms of psychological (behavioural) nature, or if physical, can be understood only using psychological concepts. (Spitzer and Endicott, 1978, p. 18)

Thus, this definition comprises three fundamental ideas within the notion of medical disorder, which altogether convey the overall message that something has gone wrong in the human organism. This gives special importance to the evaluative aspect of the concept: (1) negative consequences of the condition, (2) an inferred or identified organismic dysfunction, and (3) an implicit call for action to the medical profession, the person with the condition and the society in terms of granting exemptions from certain responsibilities to those in the sick role, as well as providing a means for delivery of medical care (Spitzer and Endicott, 1978). It is important to note that these authors’ ultimate interest is to define the concept of ‘mental disorder’, and since they decide to do this by considering it a subgenre of medical disorders, they also provide a definition of ‘medical disorder’. However, because of this ultimate interest, the definition avoids using the word ‘disease’ as, according to these authors, it generally denotes a progressive physical disorder of known physiopathology, which is not the case for most mental disorders. Therefore, the concept of organismic dys-

function, or its negative consequences, do not imply that these have a physical nature (Spitzer and Endicott, 1978). Although analysing this definition further goes beyond the interest of this revision, it is worth noting that the authors add a list of four criteria which, they argue, must be met in order for a condition to be classified as a disorder.

Wakefield's (1992) Harm Dysfunction Analysis emerges from a detailed critical analysis of several accounts, including that of Boorse (1977) and Spitzer and Endicott (1978). This author's main point is that a definition of the concept of disorder requires both an evaluation (normativist) based on social norms and a scientific (descriptive) understanding of the failure of physical or mental mechanisms to perform natural functions as designed by evolution. Thus, according to Wakefield,

a condition is a disorder if and only if (a) the condition causes harm or deprivation of benefit to the person as judged by the standards of the person's culture (the value criterion), and (b) the condition results from the inability of some internal mechanisms to perform its natural function, wherein a natural function is an effect that is part of the evolutionary explanation of the existence and structure of the mechanism (the explanatory criterion). (1992, p. 384)

For Wakefield, what follows then is a definition of mental disorders as a special case, where the nature of the cause of the symptoms determines a disorder as mental and not the nature of the symptom themselves.⁸ Therefore,

a condition is a mental disorder if and only if (a) the condition causes harm or deprivation of benefit to the person as judged by the standards of the person's culture (the value criterion), and (b) the condition results from the inability of some mental mechanisms to perform its natural function, wherein a natural function is an effect that is part of the evolutionary explanation of the existence and structure of the mental mechanism (the explanatory criterion). (Wakefield, 1992, p. 385)

However, as stated by this author, even the clearest concepts pose areas of vagueness and ambiguity, and, in this particular definition, this indeterminacy rests on how to distinguish mental from physical mechanisms (Wakefield, 1992).

All these working definitions of the concept of 'disease' share the idea that there is a discontinuity between health and disease, i.e. health and disease can be either present or absent. Nonetheless, the concept of dysfunction – that all these definitions share – admits different degrees and, therefore, raises the problem of using a continuous variable (dysfunction) as the basis for a catego-

⁸ This means that what is causing the symptom arises from a mental dysfunction and not that the symptom is mental dysfunction. Some symptoms, such as pain, are argued to be a mental phenomenon, but somatic dysfunctions may be the cause of pain, in which case, pain is not a mental disorder. Therefore, what matters regarding labelling purposes is that the nature of the cause of the symptom is mental.

rical definition.⁹ This has been described as the line-drawing problem in ‘disease’ definition (Rogers and Walker, 2017b).

Building on various disease examples (cancer, UTI, TB), Rogers and Walker (2017b) argue that the more the scientific community learns about what constitutes ‘disease’, the more difficult it is to determine the relevant dysfunction associated with a condition. As such, the absolute philosophical perspective on disease does not reflect everyday medical practice with borderline cases, drawing boundaries as necessary for decision-making and practical purposes. So, according to Walker and Rogers (2018), the concept of ‘disease’ does not seem to be classically structured since it fails to be defined in classical ways (conceptual analysis leading to exact necessary and sufficient conditions). Following from this, the authors suggest that this concept should be approached as a disjunctive and vague concept, therefore, encouraging the academic community to focus on developing specific and contextual cluster definitions for specific reasons or aims (Walker and Rogers, 2018).¹⁰

4 Conclusion: embracing complexity

By briefly revising and discussing some key approaches to the concepts of ‘health’ and ‘disease’, the indeterminateness of both concepts becomes clear, suggesting that a reflexive and open perspective towards possible specific and contextual definitions that can respond to the needs of specific quests is adopted. It also becomes clear that health outcomes and, thus, the value of a new medical intervention, will vary accordingly that depending on the conceptualisation of ‘health’ and ‘disease’. In this way, not only defining the value but also what is ‘value’ would require a contextual approach (for specific reasons or aims) that is consistent with conceptualisations of ‘health’ and ‘disease’. If, in the context of the current predominance of chronic or long-term diseases, for example, one adopts the definition of ‘health’ as the ability to adapt and self-manage (Huber *et al.*, 2011), this would introduce a radical shift in how we conceive health outcomes and, thus, how we frame the value of new medical interventions.¹¹ However, I will briefly draw on Walker’s (2019) ethical reflections about long-term treatment to reflect further on the shift introduced by contex-

⁹ “Biological functions may categorically cease altogether (the heart may stop beating, the liver stop metabolising, and the kidneys stop filtering blood), but short of absolute cessation of function, there are degrees of performance all the way up to abundantly healthy levels” (Rogers and Walker, 2017b, p. 415).

¹⁰ An example of such cluster definitions is Roger and Walker’s (2017a, p. 277) working definition of borderline diseases as “X is a disease_{odx} if there is a dysfunction that has significant risk of causing severe harm”.

¹¹ Parsons’ chapter in this publication on quality of life and life-extending treatments is a good example of this.