The Routledge Companion to Philosophy of Medicine

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The Concept of Disease

Publication details
https://www.routledgehandbooks.com/doi/10.4324/9781315720739.ch1
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Published online on: 25 Oct 2016

Accessed on: 08 Aug 2023
https://www.routledgehandbooks.com/doi/10.4324/9781315720739.ch1

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Part I
GENERAL CONCEPTS
THE CONCEPT OF DISEASE

Dominic Sisti and Arthur L. Caplan

Introduction

“What is disease?”

This simple question has vexed philosophers of medicine, yet has been overlooked entirely by bioethicists, physicians, and epidemiologists, with the exception of a rare few. On the one hand, dozens of philosophical theories paint starkly different pictures of the essential nature of disease. On the other hand, most bioethicists, policy makers, and clinicians take disease to be a given—the things we call diseases exist as such, they are bad, and they should, if possible, be treated or eliminated. Conversely, health is, quite simply, often thought to be the absence of disease.

It is across this landscape—from fine-grained analyses to straightforward pragmatism—that a multitude of perspectives on the nature and importance of the concept of disease coexist. This chapter offers a survey of this landscape and specific examples that illustrate the confusing nature and ambiguous applications of the concept of disease. We begin with a historical sketch and then examine contemporary philosophical theories of disease. To demonstrate the way these theories shape empirical realities, we offer cases across a range of medical specialties to illustrate theoretical perspectives.

From there we discuss both the clinical and health policy dimensions of the concept of disease. The concept of disease serves many purposes—identifying behavior that requires control, supporting excuses for absence from work or school, creating eligibility for benefits, and serving as the basis for exculpation in the eyes of the law. We pay particular attention to the complicated nature of psychiatric disease or disorder and lay out the significant practical stakes that depend upon the underlying theoretical orientation of disease.

Historical Thumbnails

The quest to “carve nature at its joints” and provide answers to the titular question of this chapter may be found in the metaphysics and epistemology of classifications of antiquity. Various approaches to categorizing the maladies of human existence are found in the Hippocratic Corpus and in the writings of Plato, Aristotle, and Galen (Hippocrates, 1780, Jowett, 1925, Chadwick, 1983). For the better part of two millennia, classical humoral explanations prevailed. Imbalances in the four basic substances of the body—blood, black and yellow bile, and phlegm—caused disease and disability. The medieval theological constructions of disease by
the likes of Maimonides and Aquinas both drew upon the classicists and additionally characterized various diseases in terms of moral failing (Caplan et al., 2004).

The Renaissance polymath and occultist, Paracelsus, rejected humoral models of disease and instead developed a nosology based on external substance or “poisonous” causes (Caplan et al., 1981, Porter and Rousseau, 2004). Descartes, whose anatomical studies complemented his conception of the human organism as an ensouled machine, reframed the concept of disease in a more modern way. Disease was considered a mechanical dysfunction or external attack on the body and a deviation in the teleological functioning of the automatic motions of the body.

Throughout history, the question about the nature of psychiatric disease and mental disorder preoccupied physicians and philosophers alike. Phillipe Pinel aimed to parallel his methodical systematizing of medical and mental illness with that of Hippocrates. He drew upon the philosophies of Locke and Condillac in developing a system of psychopathology that was based upon empirical observation and fact-finding (Charland, 2010). To Pinel, the various mental disorders emerged from a single kind of mental alienation. He believed that sudden, often unexpected reversals in life arising “from the pleasure of success to an overwhelming idea of failure, from a dignified state—or the belief that one occupies one—to a state of disgrace and being forgotten” could bring on “mental alienation” or mania—what we might term today depression or anxiety (Gerard, 1997).

Around the same time, an exceptionally detailed and empirically based psychiatric nosology is presented in Kant’s *Anthropology from a Pragmatic Point of View* (Kant, 2006). In *Anthropology*, Kant constructs a highly accessible nosology that was not geared for physicians but might have been useful to philosophers and individuals interested in self-treatment (Frierson, 2009). He defines two general forms of mental disorder that affect the cognitive faculty: melancholia and derangement. The first is a milder form of mental illness that is treatable, whereas the latter seems to include severe psychotic syndromes, bipolar illness, and egosyntonic disorders (Sisti, 2012).

Formal systems of post-humoral nosology find their origin in the work of Thomas Sydenham; disease classification and diagnosis slowly evolves toward what is basically the mainstream view today—an understanding that infectious agents or internal mechanisms cause localized acute or chronic dysfunctions. William Harvey’s cardiovascular findings and Morgagni’s nosology set up the foundations of understanding diseases as tissue and organ specific—a way of thinking that would prove exceptionally valuable with the later advent of germ theory (Baronov, 2008).

The 19th century saw the emergence of scientific medicine and careful observation in both empirical research and in the categorization of ailments. Claude Bernard, Rudolf Vichow, and Walter Cannon developed theories of disease based on physiological findings related to inflammation, cancerous cell growth, and the loss of homeostasis.

The renewal of interest in the concept of disease in the second half of the 20th century may have in part emerged out of a new and applied ethics and philosophy of science that used biomedicine as both a source of puzzling cases and as a professional substrate (Toulmin, 1982). Some argued that a clarification of the basic concepts of medicine—disease, illness, health, disability, etc.—would be necessary for sorting out clinical and health policy problems (Daniels, 1985).

Clinicians who examined the concepts of health and disease began to recognize deep problems in the ways clinicians equivocated in their use of these terms. Members of the first generation of biomedical ethicists sought to build a new philosophy of medicine within which a more coherent theory of disease and health served a critical role. Edmund Pellegrino, for example, wrote early nosological tracts on osseous lesions that foreshadowed over a half-century of scholarship in the philosophy of medicine and bioethics (Pellegrino et al., 1971). Pellegrino’s conceptual work on the concept of disease fit within his overall project on defining
The special, if not sacred, relationship between the virtuous physician and his patient (Pellegrino, 2008). He states that “clarification of medicine's basic concepts is as much a moral as an intellectual obligation. . . . [C]onfusion about the nature of health and disease is ultimately confusion about the concept of medicine itself” (Pellegrino, 2004).

Today there are dozens of philosophical theories on the concept of disease, from which has emerged a complex classification-of-classifications debate. The most common is to distinguish between naturalism and normativism. For example, Hofmann illustrates that across the complex array of these many accounts, theories on the concept of disease fall within two broad categories—real essence and nominal essence accounts (Hofmann, 2001). Ereshefsky helpfully adds a third category and groups the theories into the broad categories of naturalist, normativist, and hybrid (Ereshefsky, 2009). This is the tripartite division we will follow in our discussion.

**Naturalism**

Disease naturalism is the general view that the concept of disease reflects an objective reality about cell, organ, or system function or dysfunction. Physician J. G. Scadding introduced a foundational naturalistic theory grounded upon a biostatistical concept of disease. To Scadding,

> A disease is the sum of the abnormal phenomena displayed by a group of living organisms in association with a specified common characteristic or set of characteristics by which they differ from the norm for their species in such a way as to place them at a biological disadvantage.

(Scadding, 1968)

Christopher Boorse, echoing Scadding, developed and defended the most widely discussed contemporary naturalist theory. Boorse claims that the concept of disease is grounded in the autonomous framework of medical theory, a body of doctrine that describes the functioning of a healthy body, classifies various deviations from such functioning as diseases. . . . This theoretical corpus looks in every way continuous with theory in biology and other natural sciences, and [is] value-free.

(Boorse, 1975)

Disease is defined by Boorse as

> a type of internal state which is either an impairment of normal functional ability, i.e. a reduction of one or more functional abilities below typical efficiency, or a limitation on functional ability caused by environmental agents.

(Boorse, 1977)

As such, Boorse argues that diseases are recognizable against the objective backdrop of species-typical function—a concept he borrowed and refined from Scadding. Thus, the epistemic core of Boorse's theory of disease is statistical—determining species typicality is an empirical question. Boorse has labeled his particular brand of naturalism the “biostatistical theory.” Biological dysfunction is both necessary and sufficient for defining disease.

Boorse draws a distinction between the concepts of disease and illness. He defines the concept of illness as a subclass of disease: those diseases that carry with them “certain normative features reflected in the instructions of medical practice” are considered illnesses (Boorse,
To support the claim that the concept of disease is value-free, Boorse reminds us that we typically do not claim that plants or animals suffer from an illness. Rather, we describe plants and animals as simply afflicted by a disease. Potato blight, for example, renders its host diseased, not ill. One might ask, “What about cases of animal companions who are ‘sick’ or ‘ill’?” Boorse might respond that in those cases where we refer to animals, such as pets, as being sick or ill, it is in the context of a personal relationship, where we, acting as a companion and caregiver, bring to bear certain values we hold about the animal—for example, that it deserves treatment by a veterinarian.

Second, Boorse recognized that the ascription of illness grants the sufferer “special treatment and diminished moral accountability.” Thus, illness is a morally laden concept, whereas the concept of disease is, he maintains, completely value-free. According to Boorse (1975), “A disease is an illness only if it is serious enough to be incapacitating, and therefore is: (1) undesirable for its bearers; (2) a title to special treatment; and (3) a valid excuse for normally criticizable behavior” (Boorse, 1975).

Since Boorse’s theory was advanced in the 1970s, a plethora of objections have appeared. Some accuse Boorse of covert normativism by questioning the “evaluative” nature of his terminology—that the concept of function itself smuggles values because it is the product of choice on a range of things to examine (Fulford, 2001). Others deny the possibility that objective evaluations of species-typical functioning are possible.

Key objections turn on the possibility of asymptomatic individuals who present with dysfunctions or infectious agents. Are infertile individuals who do not wish to have children diseased or in any way disordered? Should individuals who have rare mutations in a suite of genes, but who are not experiencing any effects of those mutations, be considered to have a disease? Should carriers of HIV, who experience no symptoms of HIV/AIDS, be thought to have a disease (Wakefield, 2014)? Boorse has replied comprehensively and repeatedly to these and other objections (Boorse, 1997, Boorse, 2014).

Like Boorse, Lennart Nordenfelt argues that the word “disease” is simply an empirical statement and not to be taken as an evaluation of a person’s state. But, Nordenfelt’s holistic theory of health is different in the way it treats disease. Whereas Boorse works his way upward from diseases defined in terms of biostatistical deviations, Nordenfelt argues that we should work in reverse by first recognizing the suffering and the lived experience of illness and then move to examine the underlying cause of such suffering to reveal the disease state (Nordenfelt, 2007). This approach is resonant of Canguilhem’s theory of health, disease, and illness (Nordenfelt, 2007).

Clouser, Culver, and Gert provide a distinctly different account of disease, substituting the concept of “malady” for disease. According to the authors,

"a person has a malady if and only if he or she has a condition, other than a rational belief or desire, such that he or she is suffering, or at increased risk of suffering, an evil (death, pain, disability, loss of freedom or opportunity, or loss of pleasure) in the absence of a distinct sustaining cause."

(Clouser et al., 1981)

The idea here is that certain ontic evils—the authors draw explicitly on Aquinas—are objectively and universally bad. This malady account includes values but only insofar as those values are considered objective and universal; rational persons would agree that suffering, pain, injury, and death are bad and ought to be avoided. But how can an assessment of objective values be reasonably made? Who are these so-called rational persons, and how can their values be identified in a pluralistic world? Should certain maladies that predispose one to self-injury,
seizure, or hallucination—such as schizophrenia or epilepsy—but that are valued in a particular culture for spiritual reasons count as objectively evil? Clouser, Culver, and Gert’s account is very close to toppling over into normativism.

**Normativism**

The key feature of normativist theories of disease is that they all reject the possibility that objective necessary and sufficient conditions can be found to identify diseases as such. To express this position positively, normativist theories all consider the concepts of disease and health to be influenced by subjective human values to some extent. Subjective human values are beliefs, preferences, and goals that individuals or communities might reasonably hold as important to achieving their version of the good life. For a normativist, when we signify something as a disease, we are marking out something that is subjectively disvalued by society, culture, or individual preferences. Instead of appealing to purportedly objective, biologically discoverable concepts like “dysfunction,” disease signifies something that has compromised an individual’s values—i.e., some or all of those things that contribute to the well-being of a person.

Reznick, for example, argues that biological malfunction need not be involved in ascriptions of disease, and that any search for a biological cause of pathology presupposes a subjective judgment about what counts as pathological (Reznick, 1987). Such judgments may be ultimately based upon socio-political values. The normativist view on the concept of disease is now widely held. In fact, the World Health Organization’s definition of health reflects this view: “Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity” (World Health Organization, 1948).

Normativist theorists point to historical examples to make a compelling case that our concept of disease is inextricably tied to social, political, and religious values. One need only note the accepted antebellum disease of *drapetomania*—the disease that caused slaves to abscond—to see an example of how medicine and politics amplify one another through deeply flawed nosological constructs (Cartwright, 2004). Ostensibly disordered sexual behaviors—such as masturbation—are often held up as examples in support of the normativist claim that the concept of disease is subjective and in flux (Engelhardt, 1974). Homosexuality is a stark example of how moral values penetrate the scientific endeavor to categorize mental disorder and disease. It was not until 1980 that the continued medicalization of homosexuality was officially rejected by American psychiatry (Spitzer, 1981).

One response of the naturalist to these historical examples is to say they were simply wrong. We now know they were faux diseases that were artifacts of a particular ideology or historical moment. But how do we know this? Because, a naturalist might argue, we can now see they lacked the objective criteria necessary for defining disease and instead reflect bad science. Thus, blatantly erroneous historical examples actually support the naturalist cause—without an objective ideal of the concept of disease, how can we know when we were wrong about labeling something a disease?

The normative position is often found in the writings of medical and psychiatric critical theorists. Extending the position of his mentor and colleague, George Canguilhem, Michel Foucault developed a critical philosophical-historical theory on the use of medical diagnosis. He showed how the medical enterprise was a form of social control meant to cordon off individuals whose sickness or, in the case of mental illness, “unreason” rendered them useless to society (Canguilhem and Delaporte, 2000, Foucault, 2013).

Similarly, we see across the corpus of papers and books by Thomas Szasz a vociferous repudiation of his own profession’s most fundamental concept: mental illness. In his classic, *The Myth*
of Mental Illness, Szasz argues that mental disorders are whole-cloth fictions created to either marginalize eccentric behaviors or exculpate criminal acts (Szasz, 2010). In one sense, Szasz is a strict naturalist because he argues that the only legitimate mental disorders are actually diseases of the brain, which can be observed in the form of lesions or other “external” causes. In rejecting the conventional categories of mental disorder, he maintains they are indefensibly subjective. R.D. Laing also rejected the psychiatric orthodoxy arguing that mental illnesses are really just instances of mismatch and friction between an individual’s and society’s values about what constitutes the good life.

A strong normativist perspective is also reflected in scholarship on the nature of disability, where the social model of disability seems to have emerged victorious in a theoretical and political contest with the biomedical model (Sisti, 2014). The social model considers disabilities to be reflective of social and cultural barriers, and that there is nothing intrinsically dysfunctional about conditions like blindness, deafness, dwarfism, or paraplegia. Thus, it rejects naturalist definitions of dysfunction (Amundson, 1992). Although the social model has enjoyed success as reflected in landmark policies such as the Americans with Disabilities Act, some disability scholars think it has outlived its usefulness. For example, Shakespeare has argued that the social model is too blunt an instrument to provide a realistic account of disability, and that more nuanced appraisals of disability that recognize the continuum of function along which we all exist are needed (Shakespeare, 2006).

Another important political offshoot of the normative position is the ongoing examination of how ties to biomedical and pharmaceutical corporations influence the expanding broadness of the concept of disease. The basic concern here is that expert panels responsible for creating or revising nosologies often have ties to industry, and these ties influence their decisions to expand the criteria of those who are diseased or to create new diseases, which hold promise for additional profit. This latter phenomenon, now known as “disease mongering,” has been a topic of recent health policy, ethics, and sociological scholarship, though the idea that particular stakeholders endorse and motivate the creation of particular disease categories is not new (Conrad, 1975, Moynihan and Henry, 2006, Moynihan et al., 2013). Examples of purportedly manufactured or intentionally over-diagnosed diseases include mental illnesses such as depression, bipolar II, attention deficit disorder, and premenstrual dysphoric disorder (Conrad, 1975, Batstra and Frances, 2012, Chrisler and Gorman, 2015). Physical illnesses such as erectile dysfunction and high blood pressure have been singled out as examples of diagnostic expansion propelled by pharmaceutical companies’ drug sales (Carpiano, 2001). Even the most mundane of daily experiences—such as how and when one sleeps and wakes—are within the purview of medicine. Medical treatments and drugs are increasingly marketed to the public to treat “shiftwork sleep disorder” and other conditions that have been created by industrial, political, and economic structures and systems.

Hybrid Models

Efforts have been made to take the best parts of both naturalistic theories and normative theories and graft them together to form hybrid models of the concept of disease and disorder. Jerome Wakefield has developed one of the most important hybrid theories—the harmful dysfunction model (Wakefield, 1992). For a condition to be considered a disorder, it must meet two criteria. First, is the objective biological criteria defined in terms of a failure or dysfunction of an organ or body part to meet its evolutionarily determined function (Wakefield, 2007)? Second, does the dysfunction cause a particular social harm? In contrast to Boorse, biological dysfunction is necessary but not sufficient to defining disease or disorder (Wakefield, 2014).
The importance of a more holistic view of disease finds traction in George Engel’s classic papers on the biopsychosocial model of illness (Engel, 1978). He sees disease as both a biological reality—a dysfunction in the objective sense—that is fully recognized and treated only by attending to the broader context of the patient. Disease is seen as a point on a continuum of an individual’s existence. The physician who is competent in applying the biopsychosocial model would view a myocardial infarction not simply as a malfunction of the heart, but as potentially a result of the patient’s personality structure, outside stressors, environmental insults, or the result of a significant personal loss. Thus we can think of the concept of disease in Engel’s writings as something slightly different than either a natural kind or a social construction, but as a real thing that manifests within the rich context of human existence.

One of us (Caplan) has argued that a middle ground between naturalism and normativism is both possible and already intuitively exists in the way medicine is practiced. In the cases of mosaicism, hirsuteness, or albinism, one might say that there are biological abnormalities, but because there is relatively little dysfunction—correctible eyesight issues, melanoma risk for albinism—it is arguable whether these conditions ought to be considered diseases. Although there may be social harm—i.e., the significant stigma associated with being albino in many cultures leads to the condition being seen as abhorrent—the biological abnormality causes only manageable, mild impairment. Albinism is seen as unusual in many societies but not really a disease any more than a near-sighted, very light-skinned person would be viewed as diseased. To constitute a disease, both species atypicality and more than minimal dysfunction are necessary conditions (Caplan, 1992).

We might also invoke a form of pluralistic realism, such as that described by philosophers of biology in parsing the species concept, which allows for both recognition of biologically real entities and pragmatic usage of differing definitions to meet particular needs (Ereshefsky, 1992). For example, there is ongoing debate among neurologists about the definition of epilepsy, and one flashpoint is how to accurately define seizures (Walker and Kovac, 2015). It turns out that, for clinicians, the version of “seizure” that makes the most practical sense includes criteria related to outward signs and behavioral changes. This definition is of little use to basic researchers who must detect pre-ictal seizures, those not yet causing overt behavior, in electroencephalogram tracings to develop preventative medications. Such is also the case in psychotic disorders, including schizophrenia, where debate continues about the edges of clinical high-risk states, attenuated psychosis, and the fully developed syndrome (Haroun et al., 2006).

Risk states, such as high blood pressure and hypercholesterolemia, have converged with the disease they portend (Aronowitz, 2009). With new, more precise detection technologies, such as functional magnetic resonance imaging (fMRI), prodromal states of serious neurological and mental illnesses, such as Alzheimer’s and schizophrenia, are quickly changing from at-risk states into bona fide chronic illnesses. In some cases, individuals may be completely asymptomatic, but because of genetic or physiological biomarkers, risk states may be labeled as disease states (Karlawish, 2014). As such, these states must be carefully monitored, managed, and, if possible, treated lest the full-blown disease emerge. We can therefore see the concept of disease shifts according to our ability to detect and prevent diseases—the concept is a moving target tethered to the technology of the day.

**Diseases, Disorders, and the Future of Psychiatric Nosology**

As the previous examples illustrate, psychiatry presents us with some of the most challenging cases when trying to define disease. A large part of the problem is that there are, as yet, no specific biological structures or identifiable causes of mental disorders. We do know that particular
parts of the brain and neurotransmitters are implicated in some symptoms of diseases, such as schizophrenia, bipolar disorder, autism, and depression. But direct causal connections—such as we have between insulin and diabetes or fever and infection, for example—remain elusive. To some extent this is a function of definition. When a genetic cause, brain pathology, or lesion is identified as causing a mental disease—such as in the case of Rett syndrome—those disorders are redacted from the official psychiatric nosology and assigned physical disease status.

We have endorsed a hybrid model of disease, disability, and mental disorder, which recognizes that biological dysfunctions are knowable realities but that values infuse both the appraisal of those dysfunctions and the way one responds to them. In the case of mental disorder, it is not at all incoherent to argue that the function of the brain-mind is to provide the individual with both the intra- and inter-personal capabilities to flourish. How flourishing cashes out will vary from person to person, but most would agree that it would include a degree of free thought, relational abilities, independence, and the ability to discern prudential preferences (Sisti et al., 2013).

We also recognize that nosologies reflect certain practical concerns and many entities that are not “true” diseases. There may be times when application of the concept of disease is legitimate but practically pointless, such as in the labeling of conditions for which there is little understanding or cure. Other times it may make sense to call something a disease that is really not a “true” one, but it seems that medicine has the capacity to relieve the human suffering this state causes. For example, migraines may be considered a chronic disease rather than an unfortunate condition, which affords sufferers access to treatment coverage and protection under disability laws. In other words, medicalization is not always bad, and sometimes cure and treatment is possible without clarity about whether what is being treated is a disease.

One case where medicalization might be acceptable is in the description of complicated grief as a mental disorder. Although it has been simmering for decades, the debate about the status of medical grief and bereavement reached a crescendo in the run-up to the publication of the Diagnostic and Statistical Manual of Mental Disorders, 5th Edition (DSM-5)—the nosological schedule used by thousands of mental health professionals worldwide (American Psychiatric Association, 2013). It was proposed that the so-called bereavement exclusion to major depression—the stipulation that depression ought not be diagnosed within two months of a significant loss—be eliminated. This opened the door to the idea that a person could be usefully treated and medicated immediately after suffering a serious loss under the aegis of depression.

Protest ensued. On the one hand, grief or bereavement seems like a natural response to a significant loss such as the death of a spouse, child, or loved one, or the loss of worldly possessions after a natural disaster, life savings in a stock market crash, or employment (Horwitz and Wakefield, 2007). In such cases, it seemed to be stretching credulity to view grief as a part of depression. After all, are not grief and loss a normal part of life? On the other hand, serious grief would often present in ways similar to depression, it could be incapacitating, and it often responds positively to antidepressants. Why not treat it as disease in its own right?

The sustained examination of the concept of disease will continue into the future as researchers and clinicians continue to unravel causes of psychiatric disorders that are now only defined by outward behaviors. The National Institute of Mental Health (NIMH), for example, has moved to redefine the concept of mental disorder according to more granular models of genetics, neurochemistry, and neurological systems and circuitry. As previously mentioned, a diagnosis of schizophrenia may become possible in the preclinical or prodromal stages, before any obvious symptoms emerge. This brings us back to the question of asymptomatic carriers—should they be considered to be diseased and offered the entitlements of sick individuals, such as increased access to preventative treatment, social allowances, and
particular accommodations? These questions have yet to be answered but are ultimately rooted in the simple question, “What is disease?”

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Further Reading


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