Defining the boundaries of disease

Venue: Dunmore Lang College, Macquarie University
Date: 15-16 October 2015

Abstracts

Delineating disease from a naturalist point of view
Thomas Schramme

The main focus of my paper will be to discuss theoretical ways to restrict the boundaries of disease. Expansions of the concept of disease have detrimental consequences, as they might lead to the medicalization of social problems, and also because they create costs for any health care system, which provides resources for treating disease. The philosophical theory defended in my paper is usually called naturalism. It defines pathological conditions in relation to a standard of normal biological functioning.

Although such a theory has certain benefits in terms of restricting the scope of disease on the basis of scientifically accessible criteria, it also has at least one important practical drawback: To determine that a condition is a disease, according to a naturalist account, does not by itself have normative implications. In order to answer the question whether a certain pathological condition needs treatment, or ought to be treated, requires a normative assessment, which cannot be provided by naturalism alone. Yet, as I would like to argue, naturalism still has a say in providing such evaluations, because we gain knowledge about the basic elements of a good human life in virtue of developing a theory of basic biological functions. Hence, there is interplay between value-neutral (naturalist) and evaluative points of view. Altogether, naturalism can help us in setting the proper boundaries of disease and in supporting the practical determination of medical needs.

The line drawing problem
Wendy Rogers and Mary Walker

The naturalistic theories of disease developed by Boorse and Wakefield identify dysfunction as a necessary condition for the presence of disease; Boorse also considers it a sufficient condition. These accounts likewise assume that health and disease are binaries: if you have a disease you are not healthy, and vice versa. Yet, both authors acknowledge what is called the ‘line drawing problem’ – given natural variation, where do we place the line that differentiates between low or high ‘normal’ function, and dysfunction? For Wakefield, “A dysfunction exists when a person’s internal mechanisms are not able to function in the range of environments for which they were designed” (1992b, 243). For Boorse, a normal function is “a statistically typical contribution by it to their [members of reference class] survival and reproduction” (1997, 7-8). Regarding where to draw the line, he acknowledges that this is slightly arbitrary (1977, 559) but that it will be largely self evident, once function drops below a certain threshold under normal.

There are problems with both of these approaches to distinguishing function from dysfunction. Schwartz (2007, 2008) provides a comprehensive critique and offers his own solution. However, none of these authors acknowledge the inherent tension of trying to anchor a binary definition (disease/health) to a continuous variable (function/dysfunction). It
seems that most biological functions are continuous unless absent. The heart is pumping well, or not so well, or the organism is dead. Given these gradations, the line drawing problem is potentially intractable, as dysfunction does not and can not provide a clear criterion for defining disease. In this paper we explore the implications of recognising that dysfunction is a continuous variable for attempts to define disease boundaries.

**Geneticisation in the OMIM: Distinguishing Disease from Variation**

Rachel Ankeny

Prior to the genomic sequencing era, the bible for those working in clinical genetics was McKusick’s Mendelian Inheritance in Man (MIM), which appeared in multiple editions between the 1960s and the late 1990s. As indicated by its title, this catalogue was organized according to method of inheritance broadly construed, such as autosomal or X-, Y-, or mitochondrial-linked traits, and focused on phenotypes. It has now been replaced by an online version, which began in the mid-1980s, called Online Mendelian Inheritance in Man (OMIM), and which is a continuously updated catalogue which documents the molecular relationship between genetic variation and phenotypic expression. This paper explores this resource and its evolution with particular attention to how disease is distinguished from clinically-irrelevant variation, as well as how phenotypic variation continues to be captured in cases where there is no obvious genotypic association (using examples from the so-called Phenotypic Series which captures genetic heterogeneity). It is argued that the hybrid compromises encoded into OMIM given its historic roots dating to before the genomic era reflect the underlying difficulties facing the field of clinical genetics itself, namely that many ‘non-genetic’ disorders are included and many variations without clinical relevance also are considered to be within its purview. Although the ‘geneticisation’ of disease has been well-recognised in popular media and with reference to specific forms of screening programs, this example allows exploration of some of the internal epistemic, methodological, and institutional causes that continue to fuel the geneticisation of disease in contemporary medicine.

**A negotiated, sociotechnical, outcomes-oriented approach to diagnosing health-related conditions**

Stacy Carter and Chris Degeling

The problem of ‘too much medicine’ – including ‘overdiagnosis’ – underpins our interest in disease definitions and their application through diagnostic categories (Carter et al., 2015). Drawing on Hesslow (1993) and others who claim our concepts of diseases are goal-dependent we hold that, for the purposes of practicing medicine, it is most useful to conceptualise diagnostic practices and their founding categories in relation to outcomes. One way in which an outcomes-oriented approach to diagnosis is useful is as a direct counter to the potential for overdiagnosis. We contend that the worst excesses of diagnostic practice occur when the search for diseases becomes an end in itself. This obscures the proper goal of healthcare, which should be to reduce the suffering, or increase the wellbeing, of individuals and populations. Accordingly, disease categories should hinge on the potential of the resulting diagnostic practices to reduce suffering or promote wellbeing in patients and citizens, rather than on the ontological security of the definition. Shifting the focus of diagnostic thinking away from ontology and towards outcomes will be facilitated by: 1) recognising that the distinction between disease and non-disease is negotiated in sociotechnical systems; 2) taking a probabilistic, population-focused approach to the practice
of medicine; and 3) recognising that diagnosis cannot be purely descriptive: it has an inescapable normative dimension (which includes the values of the person being diagnosed). The shift away from predominantly ontological diagnosis and towards outcomes-based diagnosis changes the goal of diagnostic medicine from finding every possible case to ensuring that labels are applied only when they are likely to be good for the person being labelled. To illustrate the distinction and tension between ontological and outcomes-based diagnosis we will conclude with three examples: screening and testing for prostate cancer, diagnosing and managing latent and active tuberculosis, and diagnosing body dysmorphic disorder.


**Boundaries from Biology**

John Matthewson and Paul E. Griffiths

The precise boundaries of both the general category of disease and of particular diagnostic categories are highly contested. It is in exactly this type of situation that disagreements between commentators can degenerate into the ‘dull thud of conflicting intuitions’. Opinions regarding particular examples differ or are unclear, so we require principled, independent reasons to prefer one perspective to another. Here the method of explication can be of use – make the concept more precise through stipulation, and then justify that stipulation through reference to the utility of the new, refined concept. In this paper we look to the broader biological sciences, beyond biomedicine, to find reasons to adjudicate conceptual disagreements about disease. We identify two distinct normative criteria implicit in the basic structure of modern biology; two ways in which biologists might say an organism is doing better or worse from an objective point of view. One of these criteria regards whether the organism is functioning as designed by natural selection.

The other regards whether the organism exhibits adequate fitness relative to its peers. In philosophy of medicine these two criteria are usually thought to underwrite conflicting definitions of pathology. In contrast, we show that not only are they compatible, both are needed to properly characterise clearly distinct kinds of biological failure. To illustrate, we identify and provide examples of four ‘ways of going wrong’ from a biological perspective, each of which is frequently encountered in nature. Two of these arise when a biological structure is unable to perform its evolved function, either due to mechanistic failure (such as a faulty leptin receptor) or a novel environment (such as being surrounded by abundant high-sugar foods). The two further ways arise from constraints on fitness, either due to environmental hostility (such as a drought), or unpredictability (such as the increased risk of metabolic disease in those born during a transition from traditional to ‘westernised’ diets.)

We finish by showing how these criteria can be put to further use, as they provide a principled and independently motivated framework from which to assess specific phenotypes, etiological factors, or sequelae as one or other form of biological failure. This is illustrated by showing how the framework categorises phenotypes statistically associated with ageing as cases of either pathology or normal physiology of that life-stage.
Truth or spin? Disease definition in cancer screening
Lynette Reid

Debates over medicalization are framed in terms of the harms and benefits—and the interests at stake—in deciding whether or not to extend the category of disease to capture new phenomena. The terms of this debate may leave undisturbed our presuppositions about the nature of disease as such. In cancer screening, by contrast, the most successful screening program to date (the pap smear) medicalized a risk factor (precancerous abnormalities) and introduced treatment for this risk factor, while more controversial screening programs (breast and prostate cancer) attempt early detection of the disease itself. Screening programs designed to discover the disease itself have in turn engendered substantial and surprising uncertainty about the clinical significance of the disease entity.

Debates over the nature of disease have run along traditional philosophical fault-lines: advocates of conventionalism or normativity in disease definition face off against realist or naturalist accounts of disease. Using examples from cancer screening, I argue that realism about the biological processes underlying disease and health supports a pragmatic account of disease definition—not in the generic and sometimes anti-philosophical sense that common-sense judgments of usefulness should substitute for careful normative and scientific reasoning, but in the precise sense that the definition of disease must be crafted for the specific decision-making context of clinical medicine and the patient’s life course. This landscape includes normative and conventional factors that have robust reality and are describable and explicable by the “special” (biological and social) sciences. Hence the so-called “professional consensus” approach to disease definition is not one that asserts conventionalism against realism; rather, it recognizes the complexity of summarizing with a label what is in fact a complex decision-making process integrating probabilistic information, self-awareness of our incomplete knowledge, and the contingencies of medicine’s current ability to make a difference through disease management, requiring a careful normative balancing of harms and benefits of alternative courses of action.

Through the (related) lenses of decision-making in the context of clinical management and in the patient's life-course, I evaluate the recent proposal of an NCI working group to clarify the status of clinically ambiguous cancers and rename them (IDLE—indolent lesions of epithelial origin—is suggested). I evaluate the concern that this is a form of unacceptable manipulation of the informed consent process, and reject the criticism, arguing that the form of realism that underwrites such criticisms is simplistic.