

What is a disease?

Disease, disability and their definitions

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At first sight, the answer to “What is a disease?” is straightforward. Most of us feel we have an intuitive grasp of the idea, reaching mentally to images or memories of colds, cancer or tuberculosis. But a look through any medical dictionary soon shows that articulating a satisfactory definition of disease is surprisingly difficult. And it is not much help defining disease as the opposite of health, given that definitions of health are equally tricky. The World Health Organization’s claim that health is “a state of complete physical, mental and social well-being, not merely the absence of disease or infirmity” (WHO, 1946) has been praised for embracing a holistic viewpoint, and equally strongly condemned for being wildly utopian: the historian Robert Hughes remarked that it was “more realistic for a bovine than a human state of existence” (Hudson, 1993).

It might not be easy to articulate what a disease is, but we like to think we would at least all know when we saw one. Unfortunately, this is problematic as well. Notions of health are highly context-dependent, as human diseases only exist in relation to people, and people live in varied cultural contexts. Studies in medical anthropology and sociology have shown that whether people believe themselves to be ill varies with class, gender, ethnic group and less obvious factors such as proximity to support from family members.

What counts as a disease also changes over historical time, partly as a result of increasing expectations of health, partly due to changes in diagnostic ability, but mostly for a mixture of social and economic reasons. One example is osteoporosis, which after being officially recognized as a disease by the WHO in 1994 switched from being an unavoidable part of normal

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ageing to a pathology (WHO, 1994). This has consequences for sufferers’ sense of whether they are ‘normally old’ or ‘ill’, but more concretely for their ability to have treatment reimbursed by health service providers. Another well-known example is homosexuality, which has travelled in the opposite direction to osteoporosis, through medical territory, and out the other side. After being redefined during the nineteenth century as a state rather than an act, in the first half of the twentieth century homosexuality was viewed as an endocrine disturbance requiring hormone treatment. Later its pathological identity changed as it was re-categorized as an organic mental disorder treatable by electroshock and sometimes neurosurgery; and finally in 1974 it was officially de-pathologized, when the American Psychiatric Association removed it from the listed disease states in the Diagnostic and Statistical Manual IV (Bayer & Spitzer, 1982).

Why is it important to know what a disease or disability is? One reason is practical: because today’s medicine has an unprecedented ability to actually do things, it matters a great deal what we decide to tackle. The ability to make powerful, effective interventions into people’s health brings with it new ethical responsibilities. If we want to ensure that

limited healthcare resources are appropriately distributed, for example, we must have a reasonably clear idea, first what a disease is, and second, which diseases are most worth the investment of time and money.

More subtly, it is important to define disease because of contemporary biomedicine’s power to intervene not just in people’s health status but also in domains of their biology where the effects are morally, and economically, problematic. For example: Is someone with a genetic predisposition to a disease already ill? I may be asymptomatic but the diagnosis certainly makes a difference, not just to my future but also to my present. With a predisposition I am not actually sick (although an insurance company or employer may consider me to be), but neither am I quite the same person as I was before: getting the diagnosis may be one of the most traumatic events of my life, and may place major psychological and ethical burdens on me. So am I well, or ill? Or what?

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How do we distinguish properly between real diseases, and human behaviours or characteristics that we just happen to find disturbing? Recent discussion of this question has focused on the use of psychopharmaceuticals, and the most widely cited example is children with attention deficit hyperactivity disorder (ADHD; Zwi et al, 2000). In the past 15 years, diagnoses of

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children with ADHD have rocketed (Gottlieb, 2002), as have prescriptions for drugs to control it. Critics argue that the diagnosis of ADHD is really about badly behaved children whom parents and schools cannot control; meanwhile, proponents say that children behave badly because they have a disease that requires pharmaceutical intervention.

Are new disease entities being created to match drug development? As the business literature shows, new clinical diagnoses are often welcomed primarily as opportunities for market growth (Moynihan et al, 2002). One recent example of this is female sexual dysfunction (FSD). The huge commercial success of sildenafil (Viagra) for erectile dysfunction in men provides a strong motivation for drug companies to identify an equivalent market (that is, condition) in women. And some ethicists feel that drug companies were, to put it mildly, over-involved in the medical consensus meetings held between 1997 and 1999 that effectively drew up very inclusive clinical criteria for the definition of FSD (Moynihan, 2003).

Even in the absence of overt commercial interests, are new diseases being 'created' simply to fit the ability to diagnose them (Smith, 2002)? This is a trickier question, because of course it genuinely is the case that diseases will be poorly diagnosed until they have been properly characterized. No one would claim that if a technology allows a condition to be identified for the first time, there was no real disease before. But there are cases in which whether something is defined as 'a pathology' depends less on its effects than on whether it is consistent with a new set of medical criteria. An example here is joint hypermobility (Grahame, 1999). Being double-jointed used to be considered within the upper range of normal, and sometimes even an asset: as a spectacularly bendy little girl I did well at ballet and gymnastics, and having hypermobile fingers can be useful for pianists and flautists (Larsson et al, 1993). But joint hypermobility often accompanies heritable connective tissue disorders (HCTDs), and recent revisions of HCTD classification include hypermobility not just as a symptom of disease but as a disorder in itself

(Beighton et al, 1988; Grahame, 1992). Some of the HCTDs are relatively benign whereas others have more severe consequences, and considerable investigation (genetic and other) is needed to make a differential diagnosis. Therefore having this characteristic can now be your entry card into a world of testing. Since childhood, then, I have moved from 'enviably flexible' to 'at risk of several unpleasant disorders'; a fairly major transformation, while my everyday experience of hypermobility has not changed at all.

Defining disease is difficult, disability is worse. There are problems even with deciding where to look. Does disability lie in the person? Or somewhere else? Where does the cut-off point between physical variation and disability lie? Is there in fact a cut-off point? Until recently the only coherent model for thinking about disability was a medical one, in which disability is seen as a nominative pathology: a disease, degeneration, defect or deficit located in an individual. Exactly what constitutes disease, degeneration, defect or deficit here is decided by reference to a biomedical norm. It is therefore helpful to have a biomedical norm available, which might explain why the idea of 'disability' as a category arose in parallel with medical standardization.

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Increasing dissatisfaction with the limitations of a purely medical perspective for comprehending the whole experience of disability has generated several alternatives based on the social model (Oliver, 1996; Shakespeare & Watson, 2002). The social model's fundamental criticism of the medical model is that it wrongly locates 'the problem' of disability in biological constraints, considering it only from the point of view of the individual and neglecting the social and systemic frameworks that contribute to it. The social model distinguishes between impairment (the biological substrate, such as impaired hearing) and the disabled experience. In this view the presence of impaired hearing is one thing, while the absence of subtitling on TV is quite another, and it is the refusal of society to make the necessary accommodations

that is the real site of disability. A social model does not ignore biology, but contends that societal, economic and environmental factors are at least as important in producing disability.

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On top of this, the personal experience of disability is not always predictable, and it can be very different from the experience of disease. Most sociological models of acute and chronic disease see it as a disruption to an ongoing personal identity (Bury, 1982). In part this was confirmed by a study that I carried out together with Christoph Rehmann-Sutter and Christine Rippberger in Switzerland between 1998 and 2001, in which we compared the attitudes of potential providers and potential consumers of future somatic gene therapy (Scully et al, 2004). People with multiple sclerosis clearly identified their illness as a disruption, "something that has happened to me." Many forms of disability are also experienced as disruptions, especially those that occur in the course of a person's life as a result of ageing, trauma or illness.

But our own and other research has shown that an impairment, especially one that is congenital or genetic, and is stable rather than progressive, can also form an important part of a person's identity. In our study, some people with impairments such as genetic deafness or achondroplasia made statements like: "If you take these [disabling] elements away from me, I wouldn't be X, I would no longer be that person." Strikingly, although most participants gave their ethnic group as Swiss, more than one Deaf participant chose "Deaf culture". To locate their primary identification with other people with their disability, even above their nationality, demonstrates its importance to their sense of identity.

The example of deafness is a particularly interesting one. Many culturally Deaf (the convention is to use lowercase 'deaf' to indicate the condition of hearing impairment, and uppercase 'Deaf' to indicate the cultural

grouping) people consider themselves to be not disabled, but a linguistic minority. Although the available evidence suggests that the majority of Deaf people have no preference for having deaf or hearing children (Stern et al, 2002; Middleton et al, 2001), some clearly do, and this has already given rise to at least one high-profile case. In early 2002 a lesbian couple, both with congenital hearing impairment, used a sperm donor with a heritable form of deafness to increase their chances of having a deaf child. Note that they did not reject having a hearing child, only that they felt a deaf one would be "a special gift". The couple have so far had two children, both hearing impaired (Mundy, 2002). Public responses ranged from outrage to a defence of the couple's right, not only to have a child, but to choose the kind of child they wanted to have.

Shortly after this, an Australian newspaper reported that a deaf couple from Melbourne planned to use preimplantation genetic diagnosis (PGD) to ensure (not, like the American couple, to increase their chances) that they would have a child with normal hearing. This time there was virtually no debate of the ethical grounds for the parents' action. Because the use of PGD in Australia is restricted to preventing the transmission of disease, the local regulatory Infertility Treatment Authority was involved because "we have to ask if deafness is a disease ... Some people would say deafness is a disease. Others would say it was an unfortunate condition" (Riley, 2002). No mention was made of those who would say that deafness is neither of these, but another way of being.

I give this example not to support the right to choose hearing-impaired infants, but to illustrate that the lines drawn around

normality, abnormality and disability are not self-evident. These lines determine many moral choices in research and health-care, and they shift according to experience and perspective. For most commentators on the case, deafness is a disability and therefore, in ethical terms, a harm. For the Deaf who think of themselves as a cultural or linguistic minority, choosing deafness is more

Although this is an extreme example, similar arguments may be used for conditions that are more unequivocally disabling than deafness. For people with achondroplasia or other skeletal dysplasias, many of the disadvantages they encounter are not intrinsic to the condition but are due to society's reluctance to do things like install light switches lower down on walls; and those bits that are intrinsic, such as joint pain, are not bad enough to justify medical interventions. Like the theoretical model, these perspectives suggest that 'disability' as an experience should not be confused with simply having an impairment.

The response of a scientist to all this might reasonably be "So what?" Even if it is true that a medical model gives an inadequate account of the experience of disability, biomedical science is not concerned with disease experience, or even with ethics: its goal is the understanding of disease processes. But science does not stand above the culture in which it operates, and the influences flow both ways. It is the cultural framework that tells scientists what they should turn their attention to, and in this article I have been suggesting that biomedicine's contemporary power means that it can no longer adopt ambient ideas about disease and disability without running

into tricky areas of ambiguity and, potentially, ethical difficulties.

The opposite influence is the effect of science on everyday life. Biomedical explanations have enormous authority in today's world, and the status of genetic explanations is particularly high. There are relevant questions to be asked here about defining disease or disability in terms of the possession of a genetic marker. For one



Alison Lapper (8 months) by Marc Quinn (2000). Marble and plinth (83.5cm x 40cm x 65cm) © the artist. Courtesy of Jay Jopling/White Cube (London, UK). This sculpture caused controversy in England when it was chosen as one of two pieces to occupy the vacant fourth plinth in Trafalgar Square, London. The work will probably be installed on the plinth in the Spring of 2005 and will remain for 12–18 months.

like choosing to practice their Judaism, or to send their child to a Rudolf Steiner school: a cultural choice that closes down some options but opens up others that are equally valuable. Some Deaf people might still choose to avoid deafness in their children to protect them from social disadvantage. Others would believe that societal prejudice is not a good reason to prefer a hearing over a hearing-impaired child.

thing, a relatively small proportion of impairment is directly attributable to genetics. Most disability is caused by events that occur after birth: ageing, illness and trauma, including war, in which genetic factors may have little or no role. Nevertheless, as with disease, the ever-increasing amount of genetic information available encourages the search for genetic aetiologies for all forms of disability.

As noted earlier, one ambiguity is whether the carrier of a genetic predisposition should be considered ill or not. In addition there is a real risk that the accumulation of gene loci associated with disease leads to the conflation of the marker and what it marks. Note that this criticism does not hinge on whether the allele concerned really does cause the phenotype. There is no doubt that genetic factors are involved in illnesses and disabilities, but exactly how they interact with environmental and social factors is likely to differ for every condition. Critics of genetic determinism properly deplore the tendency to ignore non-genetic influences. The point here is a slightly different one. Two jumps are being made: from gene to phenotype, and from phenotype to experience. Irrespective of how convoluted is the relationship between genotype and phenotype, the arguments given earlier suggest that the 'harm' of the impairment is not straightforwardly related to phenotype. What ought to concern us about disease and disability is the disadvantage, pain or suffering involved, and in a sense the impairment is always a kind of surrogate marker for this experience. By defining disease or disability in terms of genetic loci, the relationship to experience is made a step more distant: removed not just from the lived experience of the phenotype, but from the development of the phenotype itself. Of course the size of this separation depends on the condition, and in many cases makes no

real difference: it would be both stupid and offensive to suggest the need to examine lived experience before deciding that having familial colon cancer entails suffering. Nevertheless, for a lot of conditions that at the moment are called disabilities, and bundled together with more easily definable diseases, the situation is not so simple.

One take-home message here is that, although disease and disability are regularly lumped together, conflating them is often misleading. Another is that science never simply reflects cultural understandings; it simultaneously helps craft the definitions as well. Choices of such mundane things as disease models and diagnostic criteria, then, are not just about research agendas or commercial influences. At their heart they embody profound ethical debates about identity, human rights and the tolerance of difference.

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doi:10.1038/sj.embor.7400195