

Scientific Contribution

The Geneticization of Diagnostics[★]

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Abstract. “Geneticization” is a term used to describe the ways in which the science of genetics is influencing society at large and medicine in particular; it has important implications for the process of diagnostics. Because genetic diagnostics produces knowledge about genetic disease and predisposition to disease, it is essentially influenced by these innovations in the disease concept. In this paper, I argue that genetic diagnostics presents new ethical challenges not because the diagnostic process or method in genetic diagnostics is ethically different in kind from traditional medical diagnostics, but because it relies on a neo-ontological concept of disease in a context of genetic reductionism. Geneticization has not produced a radically new concept of disease, however, but has introduced innovations into the classical ontological concept of disease. When this new concept of disease is held in tandem with genetic reductionism, we are led to the absurd conclusion that disease is the very essence of the human being. I argue that neither the neo-ontological concept of disease nor genetic reductionism is necessary for a proper understanding of genetic diagnostics.

Key words: concept of disease, diagnosis, diagnostics, geneticization, genetic reductionism, genetics

Introduction

“Geneticization” is a term used by Abby Lippman (1991, pp. 17–19) to describe the ways in which stories about health and disease are increasingly being told in the language of genetics. This discourse, according to Lippman, is reductionistic in its description of the human condition and increasingly takes genetics as the one conceptual model to explain health and disease. This not only directs how our resources are spent, but also has an important influence on our attitudes and values. Human biological constitution, health, disease and behavior are defined, at least in part, by the DNA code, and genetic technologies are adopted for diagnosis and treatment.

The concept of geneticization has received a fair amount of attention. Henk ten Have, for example, has carefully analyzed the idea. He argues that the concept of geneticization can be studied on several

levels: conceptual, institutional, cultural, and philosophical. He concludes that geneticization primarily operates as a heuristic tool, disclosing areas for philosophical examination and refocusing moral discussion (ten Have, 2001, pp. 299–300). Although I would not maintain that geneticization has engulfed all thinking about genetics, both in popular culture and the scientific community, the work of such authors as Lippman, ten Have, and Nelkin and Lindee (1995) shows that geneticization is a real force that deserves consideration.

This paper is an examination of the ways in which geneticization has affected the practice of medical diagnostics, a term I use to mean the theory and practice of labeling patients with a diagnosis. The biotechnology industry is flourishing as a result of genetic discovery. This industry is developing not only new treatments, but also new diagnostic procedures. Developments in the science of genetics have had a profound technical impact on the practice of diagnostics, but the social and ethical impact is equally profound. For instance, the ability to predict late-onset maladies such as Huntington’s disease can influence important life choices of those affected and their families. Knowledge of one’s

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genetic makeup can direct choice of marriage partner and choices about childbearing, adoption and use of laboratory-assisted reproduction. The dangers of social stigmatization and job discrimination because of knowledge about particular genetic conditions have been well described (Nelkin and Tancredi, 1989).

My focus, however, is primarily conceptual and philosophical. That is, I am not interested so much in the empirical question of the extent of geneticization in diagnostics, but rather in pointing out what follows conceptually from the thesis. In my estimation the ethical challenges that arise from the geneticization of diagnostics stem not from new technology, but from new ontology, the reconceived genetic understandings of the human being and of the concept of disease. I will argue that genetic diagnostics presents new ethical challenges, not because the diagnostic process or method in genetic diagnostics is ethically different from traditional medical diagnostics, but because it relies on a neo-ontological concept of disease in a context of genetic reductionism. Although this "new" ontology is not really new at all, but just a contemporary twist on a very old idea, the combination of an ontological conception of disease and genetic reductionism leads to a very strange conclusion: we *are* our diseases.

To show how this conclusion comes about, I first argue that the value considerations that are claimed to set genetic diagnostics apart are present in all medical diagnostics. Then I argue that the new ethical challenges of genetic diagnostics emerge from the new way of viewing the ontology of diseases that are taken to be genetic in origin.

Is genetic diagnostics fundamentally different from conventional diagnostics?

It might seem evident that geneticization has fundamentally changed diagnostics. Kurt Bayertz (1998), for instance, argues that molecular genetic diagnostics differs in ethically relevant ways from traditional medical diagnostics.

(1) Bayertz argues that the goals of medicine are primarily therapeutic, but that there is a gulf between the ability to diagnose genetic disease and the ability to treat it; furthermore, the gulf is not likely to disappear any time soon. The new genetics has given us the ability to diagnose and even predict the onset of diseases for which there is no treatment.

(2) According to Bayertz, it is difficult to limit genetic knowledge. It can be duplicated, transmitted, and combined in such ways that it assumes completely different characteristics. Genetic diagnoses are of interest not only to their bearers, but to families, insurers, employers, etc.

(3) Bayertz argues that the predictive aspect of genetic diagnoses is one of its most significant distinguishing characteristics. Unlike other diagnoses, genetic diagnoses can predict a disease long before the onset of symptoms. This is an attractive aspect of genetic diagnoses, but it is also problematic in that people are "placed in an ambiguous position between health and disease," and this has the potential to raise serious psychological and social problems.

(4) Related to the predictive element of genetic diagnostics is the fact that genetic diagnoses are probabilistic. Because the onset of disease depends not only on genes but also on non-genetic factors, genetic susceptibility to disease does not guarantee disease, but only a particular probability of disease. Bayertz recognizes the stakes here for the concepts of health and disease. Since every human is susceptible to some disease, genetic diagnostics, if it looks hard enough, will find that everyone has some genetic susceptibility to disease. When everyone is "diseased" in this sense, the concept of health is replaced by what he calls "universal presymptomatic multimorbidity."

(5) Finally, Bayertz argues that one of the great attractions of genetic diagnostics is the ability to establish particular risks for particular individuals. This could lead to coercive screening programs in the name of genetic health. People with a particular genetic predisposition that is subject to control by some treatment would suddenly find themselves with a new responsibility to be tested and treated, and might be blamed by society if they fail to take the steps to treat or even to prevent their own disease.

Bayertz recognizes that these problems are not in themselves sufficient reasons to reject genetic diagnostics, but they do point out the need to balance the risks and benefits of genetic diagnostics and to minimize its harms. All this is correct, and I would not want to dispute it. However, I would argue that these aspects of genetic diagnostics, with one exception, are not significantly different from other diagnostics.

(1) It is true that genetic diagnostics gives us the ability to diagnose and predict diseases for which there is no treatment. However, genetic diagnostics is not unique in this respect. For example,

non-genetic diagnostic techniques have provided the ability to diagnose many cancers and neurological diseases for which there is no adequate treatment. One might respond that there are palliative treatments that affect the course of these diseases, but the same could be said for virtually all genetic diseases. As Dorothy Nelkin (2000) points out, until the twentieth century, physicians concentrated on diagnosis because they had few effective therapies to offer. Diagnostics became a means to further knowledge about disease in general. Genetic diagnostics is now at an analogous stage in the development of medical knowledge. Hence there is no significant difference between genetic diagnostics and conventional diagnostics on this count.

(2) The problem of limiting knowledge of medical condition by duplicating, transmitting and combing bits of information is not unique to genetic diagnoses. Knowledge about sexually transmitted diseases, cancer, and a host of other conditions has broad social importance and this is not necessarily related to genetic diagnostics. The problem of privacy of medical records in the age of computers and the Internet is well recognized and goes far beyond the realm of genetic diagnostics.

(3) The predictive ability of genetic diagnostics is again not different in kind from conventional diagnostics. Many conventional diagnoses can predict a disease long before the onset of symptoms. Simple tests for intraocular pressure can predict the onset of visual field loss due to glaucoma. Very high blood pressure is predictive of stroke without itself causing any symptoms. Known risk factors for disease have multiplied rapidly, making it increasingly possible to predict disease. The majority of these risk factors are not genetic.

(4) The related fact that genetic diagnoses are probabilistic is also shared with virtually all other diagnoses. The very notion of risk factor for any disease is essentially probabilistic, and has no necessary connection with genetic diagnostics. Genetic diagnostics does give us the ability to find something potentially wrong with everyone, but again, this is not unique to genetic diagnostics. The much-discussed World Health Organization (1948) definition of health as a “state of complete physical, mental and social well-being and not merely the absence of disease or infirmity” seems to many people to so broaden the notion of health that it makes everyone diseased, or at least, not healthy. Bayertz’s argument about changing conceptions of health, however, deserves more attention. Genetic diagnoses do indeed have important implications for the concepts of health and disease, but not

because they are probabilistic. I will address this issue in the next section.

(5) Finally, we must admit the truth of the claim that the particularity of genetic diagnostics has important social implications about responsibility for taking steps to prevent disease. However, it is not only genetic diseases that have such implications. Diagnosis of infection with the HIV-virus, for example, carries many of the same implications. But these implications do not follow from the particularity of the diagnosis. All diagnoses are particular. Kazem Sadegh-Zadeh (1981) analyzes any medical diagnosis as particular to a given time and patient, and relative to a particular physician, a conceptual system, a diagnostic method, and a set of data. This is as true of non-genetic diagnoses as it is of genetic diagnoses.

How, then, is genetic diagnostics different from other diagnostics? I believe that the answer to this question is to be found in an examination of the genetic reductionism and of the concept of genetic disease that are the basis of the geneticization of diagnostics.

Genetic reductionism

Genetic reductionism is used in several different ways. Rachel Ankeny (2002) discusses several models of reducing explanations in clinical genetics to molecular genetics. She uses the example of cystic fibrosis to show the difficulty of simply reducing phenotype to genotype. In fact, research in both molecular genetics and in clinical genetics has helped to explicate causal connections between phenotype and genotype.

Thus, while some realize the complexity of the relationship between genotype and phenotype, there is another understanding that maintains that genotype is the most fundamental root of genetics. Paul Root Wolpe (1997, pp. 220–221) shows how the new genetics challenges us to rethink our concept of human nature. He points out that postmodernism has presented us with a dizzying range of possibilities for understanding the self. For many, the “genetic self” offers an answer to this state of unease. There is a popular tendency to take genetic science in an overly simplistic way, as providing the key to unlocking all the remaining mysteries of life. Yet, many cultural images lie implicit in the work of science and go unnoticed by scientists and the public at large. Scientists sometimes search for the genetic basis of social phenomena such as homosexuality, race and intelligence as

if such phenomena were bare scientific facts devoid of any cultural origins (Wolpe, 1997, pp. 213–215). As a result of geneticization, the gene is being reified by our culture in ways that are profoundly altering our view of human life itself. The gene is not only reified, but it is made to be the most important element of who we are. This is the process of genetic reductionism. It cannot help but alter the way we view disease and hence diagnostics.

Nelkin and Lindee (1995) show how pervasively this type of genetic reductionism has entered into our culture. They convincingly liken DNA in geneticized culture to a secular version of the soul. DNA gives the body life and power; it determines the true self. It appears to be immortal, giving the body everything it needs to be brought back. The gene has turned into a way of talking about “the boundaries of personhood, the nature of immortality, and the sacred meaning of life.” (Nelkin and Lindee 1995, pp. 40–42). This aspect of genetic reductionism, which they call “genetic essentialism,” has spurred increasingly intense efforts in genetic research. Nelkin and Lindee (p.16) are careful to say that the gene of popular culture is not a biological entity, even though it refers to a biological entity and derives its power from science. The gene of popular culture, on their account, is more a symbol or metaphor for personal identity, and is used particularly to explain health and disease. In the ten years since the publication of their work, however, the Human Genome Project has brought genetics even more into our everyday consciousness. As genetic science advances and expands to further occupy the attention of the public, many are losing the ability to distinguish metaphor from scientific entity. This is nothing new, however. Many people have lost the ability to recognize gravity as a theoretical construct that effectively explains a force of nature; gravity is seen as the force itself. A similar thing is happening with the gene, and with the gene there are far more serious social ramifications.

Nelkin and Lindee (1995, pp. 149–168) discuss several ways that genetic essentialism has taken hold of contemporary culture and several of their examples show the importance of genetic diagnostics for controversial social issues. Genetic constitution is important in the context of many family relationships. We can see this in the search for “birth parents” by people who were adopted in childhood; the primacy of love and care as the bond of the family gives way to the primacy of genetic relatedness. We also see it in the lengths that some go to in seeking methods of assisted reproduction

to have children that are at least partially related genetically. The desirability of reproductive cloning to some carries genetic reductionism to the ultimate degree. Serious talk of “replacing” a lost child through cloning shows the power of genetic reductionism (Robertson, 1994, pp. 8–9).

More directly related to genetic diagnostics is the practice of linking genetic information to particular diseases, conditions, and risks. Genetic analysis can label a person as a carrier of some disease and this can profoundly affect self-identity. Geneticization has led to the search for an “alcoholism gene” (Haber et al., 2005) or a “homosexuality gene” (Pillard and Bailey, 1998). This sort of genetic reductionism can influence our expectations of people, the way we educate them, and the way we blame or exonerate them for their actions. Genetic reductionism has the potential to lift any moral blame from alcoholics, but it also transforms a person who drinks too much into one whose very genetic *essence* is to drink too much.

Genetic reductionism has implications for the way we look at crime. If one’s genes cause one to commit a crime, the crime cannot be said to be voluntary. The presence of an extra Y chromosome in males, for instance, continues to be linked in much popular imagination with a tendency toward aggressive crime. This continues despite the general recognition that much of the research purporting to demonstrate this linkage has been shown to be flawed. Furthermore, even if there is some link between genetic constitution and crime, such attributions of causation are overly simplistic; even if biology does play a role in committing a crime, it does not automatically remove responsibility for the crime (Mednick and Finello, 1983). Genetic reductionism can condemn certain people to being seen as criminal in their very essence, and hence beyond any rehabilitation.

The technological advances that have enabled the explosion of genetic diagnoses are not *per se* causing the changing *mores* of contemporary culture. Such advances, rather, give fuel to the fundamentally human quest for meaning in human life and can allow some of our baser instincts to come to the surface.

The neo-ontological concept of genetic disease

There is a second factor in the geneticization of diagnostics that, in combination with genetic reductionism, leads us to our strange conclusion. This is a neo-ontological conception of genetic

disease. First, consider the metaphysics of disease in general. Precisely defining disease is notoriously difficult, but for our present purposes we can understand disease in two very different ways. Owsei Temkin (1961) has called these two conceptions “ontological” and “physiological.”

The physiological concept of disease holds that diseases are not distinct entities; rather, they are deviations from the normal physiological function of the body. The classical humoral theory of disease employs a physiological concept. The Hippocratic view takes disease to be an improper mixture of the four humors: blood, phlegm, yellow bile and black bile. Such conceptions of disease were developed, especially in the late eighteenth and nineteenth centuries, into equally grand theories. John Brown (1735–1788) held that all disease was either an excess (sthenic disease) or deficiency (asthenic disease) of the “exciting powers” that were responsible for life itself. Claude Bernard (1813–1878) rejected ontological conceptions of disease as nonsense. For Bernard, disease is just a homeostatic imbalance – a deviation from normal physiology – and this can be observed by laboratory measurement (Stempsey, 2000, pp. 73–78).

The ontological conception of disease holds that diseases are things in themselves. The disease might be an invading entity such as a bacterium or virus, or it might be an internal entity, i.e., an altered body part such as a tumor of some organ. Ontological conceptions of disease have sometimes been idealized. Thomas Sydenham (1624–1689), for instance, equated disease with a definite *species*, by which he meant a substantial form, or essence. A disease is actually a humor or miasma that enters the body and organizes itself to become a disease coincident with a specific essence. A different sort of ontological conception of disease was held by Rudolf Virchow (1821–1902) who repudiated the physiological conception of disease that he had held earlier in his life. For the later Virchow, the disease entity is an altered body part, i.e., an aggregate of cells. The pathological aggregates of cells are not the cause of disease. For Virchow, the cells *are* the disease. (Stempsey, 2000, pp. 70–73). Geneticization appears to do with genes what Virchow did with cells.

Just what makes a disease genetic is a question that does not have an easy answer. The notion that genetic diseases are those that are *caused* by problems in the genes seems intuitively plausible, but in fact it is too simplistic to do any real work. The complexity of causation in disease is generally recognized, and Kelly Smith (2001) shows how

standard accounts of causal selection fail as sufficient explanations of genetic disease. The problem is how to select genes as the primary causal factor for a disease with a highly complex etiology. Even diseases that are generally acknowledged to be genetic fail to meet standard accounts of causal selection. Take Koch’s Postulates, which historically and primarily have to do with establishing the bacterial cause of a particular disease. The bacterial cause is established when four criteria are established: (1) The bacteria must be present in every case of the disease; (2) the bacteria must be isolated from the diseased host and grown in pure culture; (3) the specific disease must be reproduced when a pure culture of the bacteria is inoculated into a healthy susceptible host; and (4) the same bacteria must be recoverable from the experimentally infected host. Obviously, the criteria concerning culture of bacteria must be modified for genetic diseases. Smith does this as follows. A pathogen is responsible for causing a disease when (1) the pathogen is always found in individuals with the disease; (2) the pathogen is never found in individuals with conditions other than the disease; and (3) the pathogen always produces the disease when introduced into healthy individuals. Now, Smith’s second criterion is too strong even for Koch’s original intent, for bacteria that are potentially pathogenic are often present in individuals without causing disease. Nonetheless, Smith’s analysis is still instructive. Cystic fibrosis, a classic example of a genetic disease, fails to meet the Koch’s postulates test because (1) the genetic anomaly is not always found in individuals with the disease; (2) it is not the case that the genetic anomaly is never found in individuals with conditions other than the disease; and (3) the genetic anomaly does not always produce the disease in otherwise healthy individuals (Smith, 2001, p. 21).

Likewise, cystic fibrosis fails Sir Austin Hill’s more complex epidemiological criteria. These criteria include strength, consistency and specificity of the correlation between causal factor and disease, temporal precedence of the cause, a dose–response gradient, a plausible cause story, coherence with other knowledge and analogy with other causal factors in similar diseases. Cystic fibrosis scores high only on temporality, plausibility and coherence. It scores negatively on consistency and specificity, and the other three criteria yield uncertainty (Smith, 2001, pp. 21–22).

Another possible causal account has to do with manipulability. Cystic fibrosis also fails the manipulability criterion, which holds that a disease is

genetic if and only if it is best controlled or prevented by manipulation of the genes. The trouble, on a practical interpretation of manipulability, is that we do not know if this criterion is true in practice for any genetic disease, and because of ethical constraints on human experimentation, we will probably never know. On an “in principle” interpretation of manipulability, the criterion might be true, but it is only trivially true when based on the “in principle” view that *all* diseases have a genetic basis (Smith, 2001, pp. 22–23).

Smith’s own epidemiological account (pp. 23–28) is admittedly minimalist, holding that if a disease is genetic, then (1) those with the gene are more likely than not to develop the disease in question, and (2) most cases of the disease in the population must be caused by the gene. This analysis is plausible, but as its author recognizes, does not help very much to classify very many diseases as genetic.

Despite this complexity, geneticization reinforces a tendency to single out genes as the ultimate specific causal factor of diseases. This is truly a conceptual revolution, and one that leads us to see “defective” genes so starkly as ultimate causal factors that we are really looking at genetic diseases through the lens of a neo-ontological conception of disease.

Eric Juengst (2000, pp. 129–137) has given the name “genetic imperialism” to the view that since genes are at the foundation of all diseases, all diseases are best conceptualized as genetic diseases. The danger of this is that it leads many to see genetic diagnostics as having more predictive power than other diagnostics. A second set of implications of the geneticization of disease is what Juengst calls “genetic contagionism.” This extends the ontological reification of diseases that was begun in earnest in the second half of the nineteenth century with Pasteur’s germ theory of disease and Virchow’s cellular pathology. As a result of this ontological conception of disease, it becomes possible to be a “carrier” of a disease without having any symptoms oneself (Juengst, 2000, pp. 136–141). Juengst thus recognizes what we have already seen in our examination of the cause of genetic disease: genes do not serve well as sole specific causes of disease. Even the diseases most solidly held to be genetic are multifactorial in origin. Hence, this neo-ontological conception of genetic disease does not serve as well as the proponents of genetic contagionism would like it to. Again, this is not to say that genetic scientists are failing to recognize the complexity of the

causality of genetic disease. It is merely to point out the conceptual problem that geneticization presents to diagnostics.

The strange conclusion and the ethical challenge of genetic diagnostics

I began by showing that genetic diagnostics is not ethically different from any other medical diagnostics with respect to its processes or the potential uses of that information. Still, we must recognize that the ethical stakes are high in the applications of genetic knowledge. This is not because of any peculiarity in the diagnostic process. It is due, rather, to the changing understanding of some of the fundamental concepts upon which genetic diagnosis is based.

When a neo-ontological conception of genetic disease is combined with the reductionistic element of geneticization, the view that the human being just is his or her genes, we get a most curious identification of disease and one’s very being (Juengst, 1995, p. 1599). If one’s identity is to have a particular genetic constitution, and one’s particular genetic constitution is in fact a genetic disease, then one’s very identity is disease. We no longer have a disease; we *are* a disease. This goes against an eminently good commonsense separation between our selves and our diseases. If one accepts genetic reductionism and an ontological conception of genetic disease, the conclusion that we are our diseases is hard to avoid. This is far more problematic than the already problematic idea, raised by Bayertz, that everyone *has* some disease. Thus, elements of geneticization lead us to serious conceptual difficulties in understanding disease, human nature, the relationship between them, and the ethical implications of this relationship.

What is the solution to this problem? The possibilities are evident. We might abandon genetic reductionism; we might abandon the ontological conception of genetic disease; or, we might abandon both. I would argue that the final option is the correct one because both genetic reductionism and the ontological conception of genetic disease are based on metaphysical mistakes.

The more controversial point is likely to be the one concerning genetic reductionism. I have been attributing this attitude to popular culture based, I believe, on good evidence. But I also believe it likely that most people would reject this reductionistic view of human nature when its

implications are made manifest. Even most philosophers with a materialistic bent would reject this kind of reductionism as inadequate to account for the complexities of human experience. It is likely that the acceptance of genetic reductionism in popular culture is rooted in a failure to consider all the implications of such a view. Often, quick fixes to certain problems appear very attractive until all the implications of the fix have been uncovered and thought through. We may now, in our culture at large, be at this stage in our thought about genetics. The solution is to help our culture think about the conceptual implications of these quick fixes and come to appreciate the hidden presuppositions that underlie them.

Likewise, the neo-ontological conception of genetic disease, which has also become a part of the geneticized medical world, seems, if not just wrong, at least unnecessary. Our language about disease is a bit slippery. Is a defective gene the cause of a disease or the disease itself? If the former seems obviously correct, consider cancer. The disease cancer might be considered the set of symptoms caused by the pathological proliferation of a cell mass, but we are just as likely to say that the cell mass itself is the disease – that is, to hold an ontological conception of disease for cancer. We might become more sensitive to the plight of suffering individuals, however, by moving away from ontological concepts of disease, which tend to emphasize localized biological parts of people rather than suffering wholes. Physiological concepts of disease, which emphasize a malfunctioning of healthy organism as a whole, are easier to reconcile with an appreciation of a person's suffering in the context of a whole life.

The controversy about concepts of disease may in fact be avoidable. It is not obvious that we even need a concept of disease. We might do just fine to adopt a stance of pragmatism about the metaphysics of disease. Our common sense may be enough to tell us what is a disease and what is not. Like the “principlists” in bioethical theory (Beauchamp and Childress, 1994), ontologists and physiologists of disease might still come to an agreement about what conditions are diseases and what conditions are not diseases without coming to an agreement on the fundamental theory of disease. It does not matter whether one abandons the neo-ontological concept of genetic disease or whether one simply becomes a pragmatic disease-concept agnostic. In either case, the prob-

lem of making the self identical with the disease is eliminated.

So, the ethical problems raised by the practice of genetic diagnostics are rooted in two concepts, both of which are unnecessary and undesirable for understanding and practicing genetic diagnostics. Even if one wants to retain genetic reductionism, however, I would suggest that an ontological concept of genetic disease is unnecessary. Jettisoning this disease concept, even while holding onto genetic reductionism, moves us away from the eddies of absurdity involved in holding that we must *be* our diseases.

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