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Editorial

When we set about creating the journal a significant factor in favour of an online format was the ability to be responsive in a rapidly changing multi-disciplinary field. That way we could be particularly 'cutting edge' and perhaps even the first outlet to provide the space for a quality discussion and analysis on a given topical subject. This illustrates a somewhat clear relation between speed, production and competition. This is important in the field in which we research given the often heard criticism of commentary and regulation lagging behind science. However the confluence of speed, production and competition is also present in the very science we are interested in and recent events would urge caution against unreflected mimicry.

In Arthur Caplan and Glenn McGee's recent column (see http://blog.bioethics.net/) they argue that Science must slow its speed. In particular they make reference to the recent controversy over Korean cloning and stem cell scientist Hwang Woo Suk. Here it seems he raced ahead to try and gain a competitive advantage in the field of stem cell research cognisant of the prohibitive nature of the US regulatory environment. Unfortunately both ethics and science appear to have suffered. As Caplan and McGee put it: "*They ran to get ahead of the world competition. Now it seems they ran so fast they fell down*". The danger may be that stem sell research suffers generally. SCR is unlikely to have any immediate benefits and so a measured co-operative approach would surely be the best way to see if it can deliver upon its myriad of promises from radical solutions to human diseases to reductions in animal experimentation.

Similarly there is virtue in a slower reflective position in bioethics and social studies of science and technology. In being responsive to rapid developments we would do well not to fall into the same trap of quality being compromised by competitiveness or the simple reproduction of naïve scientism that uncritically becomes ensconced in overly optimistic discourses of breakthrough and progress. This we hope will guide us in our relevance to present and future issues so that we can exploit the speed of online delivery and production without running so fast we fall down. Measured topical submissions welcomed.

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Genomics and the Intrinsic Value of Plants

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Abstract

In discussions on genetic engineering and plant breeding, the intrinsic value of plants and crops is used as an argument against this technology. This paper focuses on the new field of plant genomics, which, according to some, is almost the same as genetic engineering. This raises the question whether the intrinsic value of plants could also be used as an argument against plant genomics. We will discuss three reasons why plant genomics could violate the intrinsic value of plants: 1. genomics is part of biotechnology; 2. genomics equals genetic engineering; 3. plant genomics may enhance trends that lead to the instrumentalization of plants. We will conclude that in the biotic view the intrinsic value of plants is violated by plant genomics only in case of 'the genomics equals genetic engineering scenario'.

Introduction

Intrinsic value refers to the qualities of life, freedom and health[1]. During the last agricultural crisis's involving animals in Europe, like BSE and pig diseases, many groups in society criticised the policy of the government and the EU using their own version of intrinsic value. The concept is now also applied in discussions on the genetic modification of plants, where it is invoked to criticize modern biotechnology. For example, the adherents of organic agriculture[2] consider the introduction of transgenetic material in a plant as a violation of its intrinsic value.

In contrast to modern biotechnology, the new interdisciplinary field of genomics is not concerned with single genes but with the whole genome. Plant genomics offers tools, like marker assisted breeding, that may be used by classical breeding, organic breeding and modern biotechnology alike. Nevertheless some groups claim that plant genomics is almost identical to genetic engineering[3]. Does plant genomics, as the switch to the whole genome (what could be considered a holistic view on the molecular level), influence the intrinsic value of plants?

1. Intrinsic value

The concept of intrinsic value, formerly strictly reserved for humans, is only recently well established in animal ethics. The concept means that animals have an ethical status, a value of their own, independent of the instrumental value for humans. In the Netherlands the concept of intrinsic value is even incorporated in the law on the protection of animals. Without the intrinsic value of nature environmental ethics becomes a particular application of human-to- human ethics[4]. In this traditional kind of ethics the term 'intrinsic value' is used to refer to certain conscious experiences of humans, and is thus anthropocentric[5]. In this view there is a central difference between humans and non-humans: only humans have moral relevance. Everything else has instrumental value. It is a situation of us against them. In traditional ethics agriculture is morally sound, and thus also plant genomics. It cannot interfere with the intrinsic value of plants because they have no moral standing.

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According to Bryan Norton[6] this traditional ethic works so well that we do not need a distinct, non-anthropocentric environmental ethic. He argues that the interests of people in all the diverse services that nature provides are quite enough to support nature protection. But what happens to species which are not of any service, or that run against the interests of people? Norton offers lots of other reasons why these species have to be protected anyhow. He concludes that a non-anthropocentric environmental ethic is simply redundant. Against this position Warwick Fox[7] argues that it makes a huge practical difference when we grant intrinsic value to nature. In that case the burden of proof would shift from the conservationists to the people who are destroying nature. People would have to go to court seeking permission, for example, to fell trees. As a consequence people would also have to seek permission to perform all kinds of agricultural activities like cutting grass or leaves or other plant material.

With the rise of environmental ethics at the end of the sixties the term intrinsic value was also applied to the so called 'higher' animals that also have a conscious awareness because they can experience pain. The claim that nature has intrinsic value is the cornerstone of environmental ethics. In the terminology of Henk Verhoog[8] this second step in the development of the concept of intrinsic value is called zoo centric ethics because humans only need to show respect to sentient animals. Verhoog argues that in the zoo-centric approach concepts developed in the anthropocentric tradition are extended to those animals that are closest to humans. In the zoo-centric view traditional animal husbandry violates the intrinsic value of animals, but it is also in this view impossible for plant genomics to violate the intrinsic value of plants because plants are not sentient animals. The third step in the development of the concept of intrinsic value, the bio-centric view, is an enlargement of the domain of intrinsic value to all living beings. In the biotic view intrinsic value is an absolute value, without degrees, and not connected to subjective human experience. This means that all activities of traditional agriculture violate the intrinsic value of all living beings in those activities. Does the biotic view also mean that plant genomics violates the intrinsic value of plants?

2. Plant Genomics

In the Dutch research setting, genomics is defined as 'research by means of large-scale characterization of genes and gene products into the elucidation of the way genes, RNA, proteins and metabolites interact in the functioning of cells, tissues, organs and the complete organism and its environment, both in an individual or in populations of species, as well as between species'[9]. The most distinguishing issue in the Dutch definition is 'by means of large-scale characterization'. A large-scale characterization, implicitly combined with a high or reasonable speed, places the notion of 'high-throughput technologies' at the heart of genomics research. The large-scale approach also motivates the further development of bio-informatics as a means to store, analyze and interpret the large amounts of data generated. By this combination with information science, genomics may and will help to move biology from in vivo to in silico. The ultimate and highly ambitious goal of genomics is knowledge and use of 'all': the identification and structure of 'all' genes, 'all' gene products and 'all' molecules and 'all' their interactions in 'all' parts of 'all' organisms during 'all' life spans in 'all' environments[10].

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Genomics researchers[11] promise or claim to revolutionize biology and transform biological science from a largely descriptive activity into an information science[12]. In a combination of genetics and information technology, genomics will explore and exploit gene functions in living systems[13]. The knowledge that stems from genomics is thought to be useful in addressing the so-called problems of our time, such as pollution, disease and food supply. This knowledge could trigger further improvements in diagnosis, prevention and agronomic practice. Agricultural genomics may reveal what genes or combinations of genes do in plants and livestock. As genomics compiles a complete list of genes and what each of them does, the predictive power of genetic constitution on performance could increase. In this way genomics promises to have a major impact on our understanding of plant performance and also on the relevance of genes for that performance. This would help to identify in plants and livestock the variations that have high value. This could make plant breeding more efficient, but will also allow breeders to evaluate biodiversity within crops, gene banks and other stocks in a better way. Genomics could make crop growing more local and effective, as well as encourage growth of plants with combinations of genes/characters that suit the needs of farmers and farming communities.

Genomics of plant pathogens may identify the causes of plant diseases and indicate new ways of fighting them. Moreover, it can suggest strategies for minimizing the likelihood that resistance develops. Since it can show how pest organisms resist existing treatments, it may suggest new targets for novel pest management and the industrial manufacturing of such compounds. Crops that have an increased disease resistance reduce environmental impact and farming costs. Genomics will prevent pollution by reducing the use of pesticides and weed-killers in agriculture, and it may stimulate the use of plants in the cleaning up of soils contaminated with heavy metals or other undesired compounds[14].

From a positivistic perspective, high-throughput technology-based genomics could be considered the ultimate culmination of the reductionist approach to biology: an explanation of life solely in terms of interactions of genes and molecules[15]. The explanation of biological processes in molecular terms, which are in turn reducible to chemistry and physics, would be the ultimate reduction. It is therefore interesting that in the view of some, genomics will allow for a reconstitution and focus on the organism as a whole (and population and ecosystem) and bring ‘holism’ back into biology[16].

3. Intrinsic value and plant genomics

In the biotic view, plants, as living beings, have intrinsic value. We will discuss three reasons why plant genomics could violate the intrinsic value of plants: 1. genomics is part of biotechnology; 2. genomics equals genetic engineering; 3. plant genomics may enhance trends that lead to the instrumentalization of plants.

Biotechnology can be defined as 'the science and technology aimed at understanding and using living organisms or parts thereof to improve the organism for specific human uses or to make or modify a product'[17]. In this setting, many human activities should be considered part of the realm of biotechnology, and, because human use is

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central in these activities, they would not be in accord with the biotic view. Genomics is or will become an important component of 'modern' biotechnology. Some feel that it is particularly special and will lift biotechnology to a new level; others feel that it should perhaps not even be considered part of biotechnology. In the latter case the arguments of the biotic view against genetic engineering do not apply to plant genomics. Apparently, terminology is used highly interchangeably, which may be taken as an indication that genomics is still a relatively young field of science[18].

For some people genomics is the same as genetic engineering. This ‘genomics equals genetic engineering scenario’ is far from hypothetical. Both genomics and genetic engineering deal with genes, genetic material and improvements of plants. The distinction is a subtle one at most, which is easily forgotten or miscommunicated. The term 'genomics' may turn out to be a poor one[19]. The violation of the intrinsic value of plants is often used as an important argument against genetic engineering of plants. It is argued that genetic modification leads to an instrumentalization of plants. Transgenetic plants are manufactured solely for the purpose of use by humans. The consequentialist version of the biotic view does not oppose genetic engineering as such, as a morally objectionable technique, but objects to the consequences of genetic engineering[20]. According to Visser[21] the questions to ask about genetic engineering are: do transgenic plants have a life that unfolds according their nature? Is life devoid of human interference in the interest of plants? Transferring these questions to plant genomics, the main question is: does the use of plant genomics interfere with plant life in a measure that their fundamental rights are violated?

Many scientists will answer this question by stating that the concepts of genomics and genetic engineering are neither synonymous, nor mutually exclusive. Agricultural genomics will point to genes (and phenotypes) in crop plants that could be used in the improvement of the organism or its associated agronomy for specific human uses. This improvement can be achieved through marker-assisted selection. This is an anthropocentric outlook, something that is part of agriculture in general. The improvements could also be put into action through genetic engineering. In genomics, genetic engineering is likely to be of help in answering research questions. The function of a plant gene may be easier studied in microbes or model plants in order to understand better what it does. However, application of such knowledge does not imply by definition the use of genetic engineering in plant improvement. In addition, the availability of funds for genomics research is a motivation to pursue the approaches thought to be specific for genomics. Genomics could, or is expected to, generate knowledge of plants that would allow obtaining desired improvements without the need for genetic engineering. In that case genomics is helping to maintain the intrinsic value of plants from the perspective of the biotic view. In the public perception, however, the relationship between agricultural genomics and plant genetic engineering seems much less clear. This is one of the reasons why some research programmes in plant genomics already gave priority to applications without genetic engineering[22].

Agricultural genomics will have to face all the societal concerns associated with agricultural biotechnology in general, such as (over?) regulation, corporate control, ownership, distribution of profit and benefits, and safety. An important societal concern for agricultural genomics, even when excluding genetic engineering, may be

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certain aspects of regulation. The level of regulatory scrutiny currently imposed on a genetically engineered crop is high and unprecedented for any product of plant breeding. Development and costs of such regulatory requirements may have significantly negative impacts on agricultural genomics. Alternatively, when agricultural genomics ends up in the hands of a few, very large, life sciences companies, these companies could consider regulation as a protective measure for their markets and market share. In this way, companies at the forefront of genomics innovations could be tempted to use regulation as a strategy to do away with potential competitors. Such developments may have equally negative impacts on future agricultural genomics. Agricultural genomics is a costly enterprise. High throughput technologies may make a single data point relatively cheap; total costs are considerable, due to the sheer numbers involved. Private investments far outweigh public funds, supported by mergers and continuous scale-up of life science companies. This implies that a few companies may decide over genomics research targets and applications. The corporate control of biotechnology in general, and future agricultural genomics in particular, is likely to generate considerable societal concern. Despite all the promises, agenda setting in genomics seems still focused on short-term goals. These relate to conventional, high-yield industrial agriculture aimed at profit.

Societal concerns about corporate control are immediately related to issues of ownership and intellectual property (IP). Different from plant breeding, ownership in agricultural genomics is based on patenting and patent protection. Genomics research is seen as economic investment that requires return. It seems likely that the patenting frenzy of agricultural biotechnology will continue in agricultural genomics[23]. This will raise concerns about the equity, accessibility and desirability of agricultural genomics and its applications, but no concerns about intrinsic value. When agricultural genomics is seen as a high-input, high-cost, high-protected enterprise, a related legitimate concern is about the benefits of such genomics for the developing world. The developing world will face most serious problems with food supply. This concern is known as ‘the genomics divide’[24].

Genomics can be expected to contribute to a further industrialization, economization and mechanization of agricultural production. From a biotic perspective that could be regarded as undesirable because these trends enhance the instrumentalization of plants, and thus violate their intrinsic value. Particular schools in plant breeding, such as organic farming, are still debating the acceptability of molecular markers, and similar discussions can be expected on the large-scale approaches of agricultural genomics. Moreover, any impact of agricultural genomics on a further industrialization of food production may trigger uncertainties about the safety of genomics products.

Conclusions

In general, genomics, especially when presented as a 'new' technology, will encounter all the suspicions any new technology is usually confronted with. It can be considered as a next step in the process that started with the discovery of the Laws of Genetics by Mendel and the double helix structure of DNA by Watson and Crick. Because plant genomics may be used in different kinds of agriculture, plant genomics will have to face all the ethical, moral, social and technical issues associated with agriculture in

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general. Overall, there seems place and need for substantial research into the mechanisms of agenda setting in agricultural genomics research, the extent of corporate control and the diversity of society's evaluation of such issues. Policy makers and genomics researchers seem aware of the need to include society and the need to deserve a 'license to produce' and 'a license to sell', rather than to exclude society and go on[25].

It seems likely that how genomics will develop depends on the deliverables of the first years and technological progress in high-throughput technologies. Combined with mutagenesis, genomics may succeed in relieving crossing barriers and broadening the gene pool available for plant improvement without formally using genetic engineering. In society, this is likely to revive concerns about 'playing God'. Alternatively, the increased knowledge of plants and plant performance may eventually ease society's concerns about such and other changes.

From the perspective of traditional ethics and the zoo-ethical view, plant genomics does not violate the intrinsic value of plants. In the biotic view the intrinsic value of plants is violated by plant genomics only in case of 'the genomics equals genetic engineering scenario'. In all other cases plant genomics is as good or bad as traditional agriculture or organic agriculture.

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Towards a Social Contract for Genomics: Property and the Public in The 'Biotrust' Model

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Abstract

Large-scale genetics cohort studies that link genotypic and phenotypic information hold special promise for clinical medicine, but they demand long-term investment and enduring trust from human research participants. Currently, there are a handful of large-scale studies that aim to succeed where others have failed, seeking to generate significant private-sector investment while preserving long-term interest and trust of studied communities. With project planners looking for new modes of managing such complex collective endeavors, the idea of using a charitable trust structure for genomic biobanks has received increasing scholarly and policy attention. This article clarifies how thorny questions around property rights, the right to withdraw from research, access to materials, and funding might be handled within such a charitable trust structure to help produce a viable participatory framework for genomics.

Introduction

Large-scale genetics cohort studies that link genotypic and phenotypic information hold special promise for clinical medicine, but they demand long-term investment and enduring trust from human research participants. Recent experience in the United States, the UK, Iceland, Estonia, and Sweden suggests that population-level genomic studies pose particularly difficult legal and ethical challenges.[1] Indeed, many population genomics projects with great scientific promise have failed due to unanticipated controversies over the distribution of property rights, data access, risk, and benefits across different project interest groups – such as researchers, human subjects, funders, medical institutions and private sector partners.[2] Currently, there are a handful of large-scale studies that aim to succeed where others have failed, seeking to generate significant private-sector investment while preserving long-term interest and trust of studied communities.

With project planners looking for alternative governance models for genomic biobanks, the idea of using a charitable trust structure – as proposed in a 2003 *New England Journal of Medicine* article by Winickoff and Winickoff[3] – has received increasing scholarly and policy attention.[4] This literature has highlighted the potential strengths of using charitable trust law to create an ideal institutional framework, but has also identified potential problems that may hinder its implementation. Addressing these questions directly, this article clarifies and develops this "Biotrust Model" with respect to key areas of implementation, including property rights, the right to withdraw from research, access to materials, and funding. The Biotrust Model and its treatment of property interests, as explicated in this article, remains a promising framework for community-driven governance in many genomic contexts.

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I. Theoretical background: towards a new social contract for genetic biobanks

As Winickoff and Winickoff have argued elsewhere, charitable trust law may be a useful legal and institutional tool for implementing a partnership relationship between biomedical researchers and research subjects in the genomic biobanking context.[5] This idea was a response to a stream of empirical work suggesting how the existing regime of research governance has failed to manage large genomic programs.[6] These programs have co-emerged with new scientific approaches, new technologies, and a new political economy of research, all of which have helped unsettle the existing regime of research governance in particular ways. These are worth reviewing.

First, as others have pointed out, DNA sequencing technology and bioinformatics have effectively transformed human tissue into a newly decipherable source of personal health information that uniquely identifies individuals. As a result, the tissue or blood sample has become equivalent, in privacy terms, to personal data in a medical record that can be digitized and shared across computer networks, and used to discriminate.

Second, it is a characteristic of DNA that sequences are shared across family and ethnic groups. This group turn carries important consequences for research ethics and institutional design. Sequence data derived from one person's sample can implicate close family members.[7] Furthermore, research on a particular ethnic group or group of common geographical ancestry – such as those groups studied in the human haplotype projects – implicates all members of that group whether they participate directly in the research or not.[8] This fact disrupts the pre-existing ethical paradigm of research subjects as distinct autonomous individuals, introducing important notions of group autonomy and solidarity that have emerged with force in recent population genomic projects.[9]

Third, and less noted, the creation of large genomic assemblages – often incorporating human tissue, medical information, and genealogical information -- for unforeseen research protocols has distanced genomics from the paradigm of research for which informed consent and IRB review was specifically tailored. In the past, research involving human subjects has been limited in time, and defined for a specific scientific study. Expanding the time-scale and openness of research use poses problems for achieving meaningful informed consent.[10]

Fourth, the new role of the private sector in these projects, and an increasing emphasis on property, has destabilized understandings between human research participants and academic researchers. Traditionally, this relationship involved the exchange of altruistic donation of time, bodily tissue, and medical information from human subjects in return for possible biomedical progress with general benefit.[11] However, the growing role of private industry in biomedical research and discovery has introduced property, profit and distributional struggles into the equation. Furthermore, the commodification of biological information[12] has raised new controversies around intellectual property, rights in samples, and scientific control.[13]

These trends make the design of an appropriate regulatory regime and institutional structure for genomic biobanks a novel challenge. Together they highlight how genomic research is not only a complex scientific endeavor, but also a complex social

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one. The off-the-rack regime of bioethics will no longer suffice, and instead we must rethink the processes and structures through which the affected communities and the public may deliberate upon, constitute, and enforce a new social contract for genomic research.

II. The Biotrust Model: clarifying the legal and institutional structure

The sustainability of large-cohort genomics will require institutional, procedural, and substantive legitimacy in order to secure ongoing funding, a progressive research program, and the willing participation of volunteer subjects over time. Using traditional informed consent and expert ethical review as a foundation, the Biotrust Model attempts to address these novel challenges by focusing new attention on issues of governance, constitutional powers, control of resources, and public benefit. The Biotrust Model aims to create a flexible institutional space through which the social contract around particular projects may be negotiated, ratified, and implemented. The hope is that the idea can be adapted and provide utility for new genomic projects as they emerge, especially projects attempting to implement innovative strategies for managing genomic resources for public benefit.

The Biotrust Model consists of a legal structure for handling the property rights and management of donated genetic and informational resources, and a social structure aimed at bolstering community participation, representation and trust in genomic governance – necessary conditions for sustainable collaborations. Because there has been some confusion and misstatement of how this model could operate, we believe it would be useful to clarify the model and respond directly to productive criticisms that have emerged regarding its operation.

The core idea of the Biotrust Model is to use the charitable trust as a legal framework to manage genomic resources and to govern genomic research more justly.[14] A trust is a formal legal institution in which a property interest is held by one person or set of persons (the trustees) at the request of another (the settlor) for the benefit of a third party (the beneficiary).[15] The property interest is conveyed to the trustee in a trust instrument that must clearly express the wish to create a trust. The settlor appoints a trustee of the property, who has legal fiduciary duties to keep or use the property for the beneficiary, creating a unique protective regime of trust law for safeguarding the interests of donors and other beneficiaries.[16] The creation of a trust establishes a fiduciary relationship in which a trustee holds title to property, subject to an equitable obligation to keep or use the property for the benefit of the beneficiary.[17]

To be classified as a charitable trust, the purpose must be “charitable” and aim at the public good.[18] Courts have defined “charitable” broadly in order to encourage “experiments to which it would be improper to devote the public funds or that the public would be unwilling to support until convinced by proof of their success.”[19] Allocating benefits of the trust to specific groups rather than general public might be desired in certain biobanking situations because of the composition of the donor group, because of some heightened need of a particular segment of the public, or because research results may be relevant only to a small section of the public. Directing benefit to certain segments of the public would not jeopardize charitable status of a trust. In fact, at least in the Anglo-American tradition, the charitable classification of trusts requires that the purpose involve benefiting a *class* of persons and not simply the community at large.[20]

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The charitable trust model combines a series of individual trust instruments, in which donors give certain property interests to the same trustee, the Biotrust Foundation, a non-profit organization that holds and manages the biorepository in accordance with the stated charitable purpose. The charitable trust structure would put a legally binding fiduciary obligation on the trustee to faithfully manage the resource according to the charitable purpose and the public benefit defined in the trust instrument. This fiduciary duty forbids trustees from self-dealing with the trust assets, engaging in conflict of interest transactions, and entering into transactions adverse to the trust.[21] The Biotrust Foundation would be managed according to its by-laws, which the donors would also agree to, and which define the charitable purpose and the terms of public benefit. Procedural details could be changed according to the needs of the foundation's goals, by amending its by-laws.

The Biotrust Model adds important governance mechanisms to this basic charitable trust framework. In this model, the by-laws would specify that use of the trust property would be contingent on review and approval of two bodies, the Ethical Review Committee (ERC) and the Donor Advisory Committee (DAC).[22] The ERC of the biorepository would provide peer review and ethical analysis of research protocols that call for access to biobank materials. This committee would be roughly equivalent to an Institutional Review Board (IRB), except that it would be more directly responsive to the collective interests of the donor group. Ideally, it would involve a significant number of donors so as to be more representative of the research subject population than current regulations require, at least in the United States.[23]

The Donor Advisory Committee would be a body composed of direct representatives of the donor group, and this group would help assure that the public value of the collected charitable donations would be maximized. This body would approve research protocols, but would also serve as a conduit between the donor group, the trustees, and the researchers in order to address controversial projects or issues as they arise. The DAC would provide an important democratic element to the governance of the trust, but is envisioned also as a flexible mechanism through which communication and learning could take place among the biobank constituents. In the United States, a similar body has emerged out of the Framingham Heart Study in Massachusetts, an ongoing study of fifty years in which the subject group has taken an interest and role in project decision-making.[24] These representative could be elected periodically through proxy voting, in a process akin to the election of board members by shareholders of a corporation.

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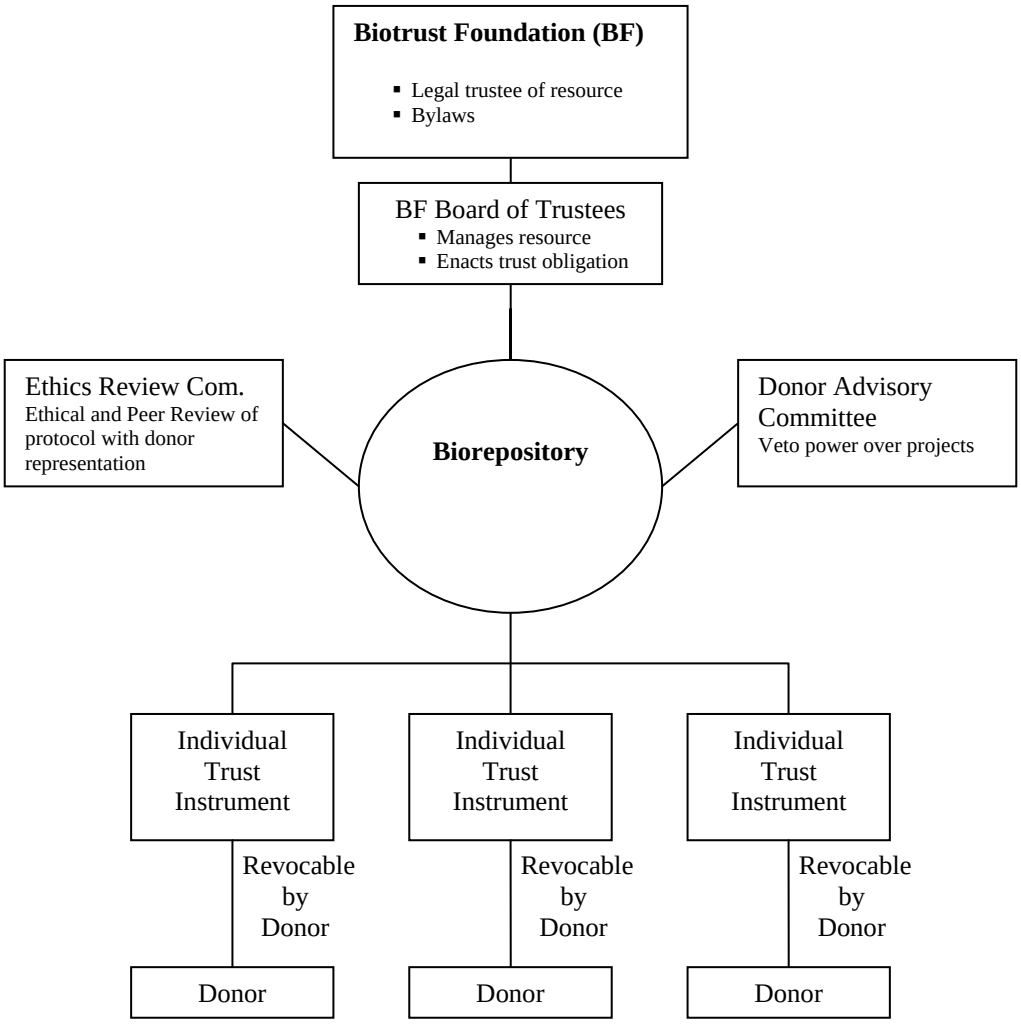

III. Response to criticisms

Critics in Europe and the UK have highlighted certain advantages of this model. First, it respects the altruistic intent of donors, while ensuring that their goodwill is not exploited. Second, it imposes a duty on the resource managers to make the resource productive. Third, fiduciary law addresses a power imbalance between the settlor/beneficiaries and the trustee, in contrast to the consent model, which has often been criticized for failing to take into account the power imbalance between doctor and patient.[25] Fourth, the separation of storage and usage reduces the conflicts of interest in making prioritization decisions about the resource, enhances the opportunity for ethical review, and encourages interest groups to participate in decision-making.[26] Fifth, the procedural mechanisms and structures are likely to help mediate among diverse interests implicated by the research. Other advantages in terms of transparency, donor representation, autonomy, and scientific utility are discussed in the original article, and need not be rehearsed here.[27]

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Recent scholarship has also raised important concerns with the legal architecture of the charitable trust model. This criticism, especially that of Boggio,[28] has been helpful as an analytical spur towards better specifying and clarifying how the model could be useful. Here we discuss the significant issues for genomic governance raised by Boggio and others, and explain how the charitable trust model could be designed to resolve them.

A. Property rights of biological samples

One concern Boggio raises is that the charitable trust idea fails to answer important questions concerning ownership of tissue samples, derived data, and the database itself. Boggio notes that "property in the body" is ethically controversial and legally inadmissible in certain jurisdictions, and this might make the model incompatible with many legal systems to the extent that it requires a formal recognition of property in the body. The question of ownership of samples is a thorny one, and it is important to clarify why establishing the charitable trust would violate neither the spirit nor the letter of this prohibition.

As a threshold issue, it should be pointed out that the charitable trust model does not require the formal recognition of "property in the body," at least not as that phrase is normally understood. The rejection of "property in the body" in certain jurisdictions limits the free alienability of bodily entities integral to personhood, and is based on the visceral and ethical disdain for commodification of the human body.[29] However, recognizing the existence of property-like interests in human tissue is not tantamount to endorsing a full spectrum of alienable property rights, for example the right to sell tissue at any time for cash compensation. As one commentator has put it,

> *. . . the equation of any property right with the full spectrum results in the erroneous impression that recognizing the existence of property rights in human tissue is tantamount to endorsing a right to sell any body party, at any time, for cash compensation . . . . A richer and more complete understanding of property rights, however, emphasizes the tremendous variety of possible property regimes in human biological materials. Property is a flexible concept, not an all-or-nothing one.*[30]

The creation of a charitable trust would not require a general property right in *the* body, but something much narrower: the recognition that personal rights of control, and use, and access in pieces that can be extracted without harm (indisputably held by the person prior to donation) may form the basis of a legal trust.[31]

In fact, the charitable trust is a legal tool for effecting this norm of non-commodification. The structure relies on the recognition of a property-like interest in donated materials only for the narrow purposes of creating an enforceable trust relationship, one that embeds control of tissue in a managed network of non-commodity exchange: samples must be used according to the terms of the trust, and the trustee enforces this use. Furthermore, the donor retains some control over the use of the donation because she can withdraw according to the trust agreement. Using a revocable trust relationship actually ensures that donors retain an equitable interest in their donation, suggesting a sort of joint control with the trust (see below).[32] This joint control of the donation does not defeat the ability to create a trust. Here the land

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analogy helps: a person can give a mortgaged property to be held in trust even though they do not own a complete inalienable right to that property.

Within some jurisdictions, such as the United States, body parts are already understood as subject to "property interests" or "quasi-property" in certain contexts, especially where the bodily materials are not integral to the person's health and functioning.[33] The idea of a limited or quasi-property right actually comports with the WHO Regional Office report that Boggio cites, recommending that participants in biomedical research should have the "primary control [of] samples or the information generates from them," and that their legal interest "is akin to a property right."[34] In these jurisdictions, all that is required to create a trust is that donors transfer these rights to the charitable trust.

Boggio is also concerned that the charitable trust model is in conflict with policies that provide that the donation of the tissue sample does not transfer property in the sample to the recipient, noting that the "Icelandic Acts on Biobanks explicitly provides that the biobanker is not to be considered the owner of the biological sample."[35] We would like to draw a distinction between ownership, which suggests the full and undivided bundle of rights associated with property, and possessing certain property interests. As Harvard property scholar Joseph Singer puts it, "the core image of ownership is ownership of a home. The core conception is the notion of absolute control; ownership is the ability to do what you like with your own, without having to account to anyone else for your actions."[36] If ownership implies absolute control and complete freedom to use, access, sell without account to others, than the trustees can't be said to "own" the samples under the charitable trust model. We imagine a so-called "donor-advised trust," in which the materials are actually co-managed by donors, trustees, and the sitting ethics board in accordance to the charitable intent of the donors. In this sense, the trustee actively holds many of the traditional "sticks" in the bundle of rights associated with property except the right to sell freely. These include rights granting access, use, and control, but only for certain limited purposes set out in the trust purpose and by-laws. Furthermore, the donors retain the ability to enforce those limitations through participatory governance and the right to revoke the gift.[37] Thus, this system is specifically designed to address the ethical, legal and social problems associated with the free alienation of bodily materials: instead of allowing donated materials to disappear into the unaccountable vortex of "research" or the market exchange, the charitable trust actually institutionalizes the connection between body part and person.

In practice, to use the "no property in the body" idea to deny research donors the ability to put these materials in a charitable trust would achieve the perverse result of facilitating their commodification. Whether the law recognizes this fact or not, biological samples have become commodified in the political economy of genetics research.[38] The Biotrust Model proposes a way to govern and regulate the exchange of bodily material in order to mitigate some of the concerns that are presented in either a system of complete ownership or complete lack of ownership. Indeed, this institutional model is specifically designed to render the exchange of human research materials more accountable to the donors and more consistent with their charitable intent.

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B. Property rights in derived data and databases

Dealing with derived data and databasing from a property perspective are less difficult to address. If the charitable trust institution itself performs research and in the process derives data from peoples' donations, then that data is clearly controlled by the trust itself, and would then have to be managed by the trustee according to the charitable purpose and by laws of the trust. The database itself, if constructed by the Biotrust Foundation, will also be owned and controlled by the trust itself. This accords with the typical treatment of data and research tools created through research.

Of course, commercial entities that use donated research material to conduct research have a legitimate interest in the fruit of their labor.[39] If the trustees decide to grant outside researchers or companies access to particular data or samples, and those research entities construct their own electronic database, ownership of this database would be negotiated in the licensing agreement between the trust and those researchers. However, based on the charitable trust model the researchers would never have complete ownership of the research material. For example, the research material could not be used for some alternative research that is different from the original research protocol approved by the trust's Ethical Review Committee and the Donor Advisory Committee. The flexible nature of the model allows the Biotrust Foundation's bylaws to determine the interest that commercial entities have over the research material. Therefore, the procedural details about the right for commercial entities to access and use research material will vary depending on the charitable goals and by-laws of the Biotrust Foundation.

C. Managing access to samples and information

As the previous sections suggest, the Biotrust Model assigns the role of managing access to the board of trustees, although in our Biotrust Model they are constrained by the Ethical Review Committee (ERC), Donor Advisory Committee (DAC) and donor rights of withdrawal (described below). This much has been discussed briefly elsewhere.[40] Boggio correctly points out, however, that our structure does not specify *how* access to samples and sensitive data will be managed by the trustees. He raises a series of questions that will help us clarify how a charitable biotrust might be implemented.

First, Boggio notes that the charitable trust structure does not answer whether and how external research groups will access samples, data derived from them, and/or sensitive health information collected along with samples. It is true that a charitable trust structure by itself does not provide an answer. But this fact provides flexibility that is actually a strength of the legal structure.[41] It will be up to the project organizers, donor group, and board of trustees to develop acceptable by-laws for the trust that may dictate guidelines for the degree of access. We would suggest that these be negotiated before the architecture is set up, perhaps through a process of public consultation,[42] but the by-laws could be written to permit later refinement in the interest of pragmatism. These by-laws, negotiated before hand and refined later, should also address any principles of prioritization as between commercial and academic protocols, another open issue Boggio has raised.

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If past experience is any judge, researchers in both the commercial and non-profit sectors will likely ask for access to biological samples, DNA sequence information, and any data that the charitable trust might hold. However, recall that under our biotrust model, the protocol and any commercial deals would have to be approved not only by the trustees, but also by the ERC and the DAC. Furthermore, access would be limited by any statutory or regulatory requirements. Privacy rules and human research protections in many countries will likely prevent the transfer of identifiers along with samples and health information, at least without individual authorization.[43]

Finally, Boggio asks whether participants would be informed if research findings might affect their individual care. This is an important issue, but one that can be dealt with through the individual donor agreements with the trust. In the interest of enhancing the autonomy of individual donors, and potential direct benefits of the research, we would agree with previous commentators that donors be able to indicate on the consent form – by checking a box – if they would like to be recontacted (so long as identifiers still allow it) in therapeutic situation.[44] Recontact could be pursued through the donor's primary care physician.

Boggio states that "in the end, the trustees will be asked to make these sorts of judgments in adopting the policies that regulate third-party access. Most of the answers will only lie in those rules governing the biobank and its contractual relations with external actors rather than in the governance framework."[45] While this could be true if the trust instrument gives trustees control, the choices of managers can be constrained and regulated in a number of ways. Additionally, we have suggested a set of policies to help guide the thinking of project planners as they consider how they might accomplish different access goals through the by-laws of the trust.

D. Implementing the right of withdrawal

As we have discussed elsewhere, a charitable trust in the biorepository context must make special accommodation for research participants, allowing them to be able to withdraw their samples from the biobank at any time.[46] Boggio raises the question of whether a revocable charitable trust is able to effectuate this right of withdrawal.

First, the right to withdraw one's samples from the charitable biorepository needs to be distinguished from the opt-out window, another mechanism previously described through which the so-called "open consent" problem is mitigated. For efficiency reasons, most biobanks seek an open permission from donors at a single point in time for future research projects instead of recontacting and reconsenting donors when new protocols arrive. These so-called "open consents" pose problems for traditional notions of informed consent, which require that the research participant understand the risks and benefits of particular research protocols. Stanford University bioethicist Henry T. Greely and others have argued persuasively that biobanks' requests for general permission should be allowed only if additional safeguards are in place.[47]

The "opt-out window" is a mechanism that increases the autonomy of research subjects who give open permission, for it allows the donors to review particular protocols and gives them a time-limited window in which to opt-out of that particular protocol. This mechanism honors the traditional norm of granting research subjects the right to withdraw from particular research project, while not overburdening the

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biobank with recontact and reconsent. Through electronic newsletters, normal mail, and a web site, the biotrust foundation would convey information to donors, allowing donors to make informed decisions about withdrawing from specific research projects for a short period of time before the research begins.

Second, the revocable charitable trust is indeed a mechanism that could effectuate the right to withdraw one's sample from the biobank at will or in cases of non-adherence of trustees to the charitable purpose. A revocable trust is a trust that allows the settlor to revoke the trust property, according to the terms of the trust instrument. Boggio states that "technically it is not a 'revocable' trust because in this case, the withdrawal of biological material of a single settler/donor does not revoke the whole trust."[48] It is incorrect as a matter of law that the creation of a revocable trust relationship between individual donors and a single trustee would require that if one donor withdraws, the entire biobank be revoked. For instance, if one made a conditional donation of land to a charitable trust, say the National Land Trust, and the conditions were not met, the property would revert to the donor without interfering with trust's other obligations. While legal partnerships might embody this "all-for-one-one-for-all" idea, revocable trusts do not. The key conceptual point is that people donate into the existing trust, but under certain conditions, such that the Biotrust Foundation has individual revocable trust relationships with each donor.

Individual revocable trust relationships are ideal for implementing the Biotrust Model that we advocate for biobanks. In a revocable trust the settlor maintains equitable rights in the trust property, though the trustee holds legal title. If a trust is not revocable the settlor usually has no equitable claim to enforce the terms of the trust or to terminate the trust.[49] Most jurisdictions assume that a trust is revocable unless the trust instrument indicates otherwise.

Another concern Boggio raises is the status of the donated material at the individual donor's death. One simple solution is for charitable biotrusts to allow for individual variations in the trust instruments that indicate the settlor's desire upon death. For example, settlors could indicate that they want the donation destroyed at their death or they could indicate that they want the sample permanently donated without any restriction.

The trust bylaws and individual trust instruments would set the policy for withdrawal. As Boggio states, withdrawal might entail a number of actions, such as returning samples to the participants, destroying samples, anonymization, removing identifying information, destroying genetic data derived from the sample or simply no longer use them. There are some concerns about allowing donors to withdraw from scientific research at any point because scientist may not want to invest in research that could possibly be terminated or damaged by a withdrawal. Pragmatism requires that once a research project is started, the donor would not have the ability to withdraw from that specific study. But at any time the donor could completely withdrawal their material from any future research.

Creating a revocable trust ensures that donors maintain the legal rights necessary to withdraw their donation and enforce the duties of the trustees. In this way, the revocable status of donations helps advance the representation, deliberation, and

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accountability of researchers to the donors, which is appropriate in research using biological material and information.

E. Funding

As stated in previous work, initial funding for a Biotrust should be committed in the public interest by state or charitable donors, and experience indicates that state governments, medical charities, and even particular disease groups may be willing seed such an entity.[50] The key benefit of the charitable trust is that the collection must be managed for public benefit. However, some relationship with the private sector may be desirable to promote research and to funnel money back to support the trust infrastructure. Once the charitable biotrust foundation is started it will likely sustain itself by charging reasonable fees for access to cover operation expenses. Furthermore, depending on the goals of the charitable trust, the biotrust foundation could contract for intellectual property rights in the research.

Boggio has stated that the Winickoff & Winickoff model insufficiently details how it could balance openness and public-benefit with commercial collaboration.[51] Boggio is correct that the Biotrust idea by itself says little about how an appropriate balance should be struck between maintaining open access and encouraging private collaboration. The optimal balance cannot be prescribed, but will have to be worked out in practice. The fact is, there are various ways that charitable biotrusts could strike this balance, depending on the goals and purposes of biotrust and its donors.

Managing the public-private interface in genomics in a way that is acceptable to those involved may be one of the greatest challenges facing these endeavors. A key strength of the Biotrust Model lies in its governance architecture -- in which power is shared across the board of trustees, donor representatives, and the ethics committee. In its ideal form, this architecture would help foster a Habermasian space[52] for public deliberation and learning not only about the use and operation of the biobank, but also about the new genetics and its effect on the political economy of health. Joint governance creates the potential ability to deliberate policies regarding the private sector. We imagine that the donor representatives will be in contact with the donor community through newsletters, public hearings and comment periods. These efforts may be useful to achieve pragmatic policies, so long as they are ratified by the community.

VI. Conclusion

Biomedical research has shifted meanings with the advent of new genetic technologies and an expanded role for the private sector. Large genomic assemblages embody these changes, but they retain a strong public character and mandate due to the collective demands of this type of research. The ongoing project to construct a genomic governance that acceptably orders the interface between public and private will likely fail until we reconceive genomics as an enterprise driven not by profit, but by collective political will. The Biotrust Model attempts to create a framework for this reconception. It is an attempt to create a vigorous public space through which a new social contract for biomedical research may be negotiated and ratified. At the same time, it seeks the elusive balance between respecting the dignity of human persons and generating public value, a balance that has been unsettled by the new

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modalities of biological science, technology, and property. In order to accomplish these tasks, it constructs a hybrid legal identity for genomic resources, one that stakes out a position between personhood and property, gift and commodity, group and individual, public and private. Its merit, if it has any, will be measured not by its theoretical novelty, but by its practical ability to open pathways of democratic governance through complex technoscientific endeavors.

Acknowledgements

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[1] See, e.g., G. Williams, Bioethics and Large-scale Biobanking: Individualistic Ethics and Collective Projects. Genomics, Society and Policy 2005; 1,2: 50–66; D. Winickoff, Governing Population Genomics: Law, Bioethics, and Biopolitics in Three Case Studies. Jurimetrics 2003; 43: 206.

[2] See, e.g., K. Philipkoski, Framingham Gene Project Killed. Wired 2001 (Jan. 2); H. Rose, An Ethical Dilemma: The Rise and Fall of UmanGenomics – the Model Biotech Company? Nature 2004; 425: 123-124; A. Abbott. Icelandic Databases Shelved as Court Judges Privacy in Peril. Nature 2004; 429: 118; J. Burgermeister. Estonia genome project lives on. The Scientist 2004 (28 April).

[3] D. Winickoff and R. Winickoff, The Charitable Trust as a Model for Genomic Biobanks. New England Journal of Medicine 2003; 349: 1180-1184.

[4] J. Bovenberg, Whose Tissue is it Anyway? Nature Biotechnology 2005; 23: 929-933; A. Boggio, Charitable Trust and Human Genetic Databases: The Way Forward? Genomics, Society and Policy 2005; 1: 41-49; PropEur Workshop on Property in the Human Genome, “Benefit-sharing and the Charitable Trust as Models of Regulation in Intellectual Property Rights” (8-9th July 2004, Cardiff University, UK). http://www.propeur.bham.ac.uk/Cardiff%20workshop.htm; In the United States, the author (DW) has been working with the U.S. Veterans Administration and the University of Utah to apply aspects of the Charitable Trust Model for governing genomic biobanking projects.

[5] Winickoff & Winickoff, op. cit. note 3.

[6] For instance, Reardon has shown how in the Human Genome Diversity Project the project planners failed to find an acceptable solution to the problem of obtaining group consent among the indigenous communities to be sampled. J. Reardon, The Human Genome Diversity Project: A Case Study in Coproduction. Social Studies of Science 2001; 31: 357-8. Also, the Framingham Heart Study population fell apart due to disagreements between private funders, university sponsors, and government agencies over data sharing and intellectual property. See Philipkoski, op. cit. note 2.

[7] This fact has, among other things, recently changed the policy discourse on DNA databanks for forensic purposes which, because of familial searching, effectively expands these databases exponentially. F. Bieber, C. Brenner, and D. Lazer, “Data mining the family tree: identification of relatives using genetic kinship analysis of DNA databases.” Conference presentation at the National Conference for Digital Government Research, Atlanta, 2005.

[8] G.J. Annas, Rules for Research on Human Genetic Variation – Lessons from Iceland. New England Journal of Medicine 2000; 342: 1830-1833.

[9] B. Knoppers and R. Chadwick, Human Genetic Research: Emerging Trends in Ethics. Nature Reviews: Genetics 2005; 6: 75-79. H.T. Greely, The Control of Genetic Research: Involving the "Groups Between." Houston Law Rev 1997; 33: 1397-430; Reardon, op. cit. note 6.

[10] National Bioethics Advisory Commission (U.S.). Research Involving Human Biological Materials: Ethical Issues and Policy Guidance (1999); 59-61.

[11] See, e.g., Richard Titmuss. 1997. The Gift Relationship: From Human Blood to Social Policy. 2nd ed. A. Oakley and J. Ashton, eds. New Press.

[12] Rose, op. cit. note 2.

[13] See, e.g., K. Sunder Rajan, Genomic Capital: Public Cultures and Market Logics of Corporate Biotechnology. Science as Culture 2003; 12,1: 87-121.

[14] Winickoff & Winickoff, op. cit. note 3.

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[15] Black's Law Dictionary, 7th Edition.

[16] For a more in depth look at trust law as a protective regime for safeguarding the interest of investors see J. Langbein, The Secret Life of the Trust: The Trust as an Instrument of Commerce. Yale Law Journal 1997; 107: 182.

[17] Most basically, charitable trust status imposes fiduciary duties on holder of property – in this case, the charity-recipient of the donor's gift. See generally Restatement (Third) of Trusts. 2003. § 5 (distinguishing trust relationship from contracts, conditions, and other arrangements).

[18] G.G. Bogert, G.T. Bogert. 1992. The Law of Trusts and Trustees. 2nd ed. 323-9.

[19] A.W. Scott. 1989. Trusts. 4rth Ed. §375.

[20] Id. at §374.7. Thanks to one of our anonymous reviewers, who astutely helped us identify, clarify, and sharpen this issue.

[21] J. Langbein, The Contractarian Basis of the Law of Trusts, Yale Law Journal 1995; 105: 655.

[22] This would effectively impart veto power over particular projects to both committees, which are envisioned to operate through majority voting.

[23] In the U.S., the so-called "Common Rule" that protects human research subjects in federally funded research requires only that "each IRB shall include at least one member who is not otherwise affiliated with the institution and who is not part of the immediate family of a person who is affiliated with the institution." 45 C.F.R. §46.107(d), IRB membership. We would recommend that the number of community members or research participants on the biobank's ethical review committee amount to half, or nearly half, of the body.

[24] Personal communication with D. Levy, Director of the Framingham Heart Study. See also D. Levy and S. Brink. 2005. A Change of Heart: How the People of Framingham, Massachusetts, Helped Unravel the Mysteries of Cardiovascular Disease. Knopf.

[25] Remarks by D. Dickenson, PropEur Workshop on Property in the Human Genome, Benefit-sharing and the Charitable Trust as Models of Regulation in Intellectual Property Rights (8-9th July 2004, Cardiff University, UK). http://www.propeur.bham.ac.uk/Cardiff%20workshop.htm

[26] Boggio, op. cit. note 4.

[27] Winickoff & Winickoff, op. cit. note 3.

[28] Boggio, op. cit. note 4.

[29] The moral fabric of western society is deeply woven with strains of thought condemning the characterization of the human body and its derivatives as property that can be sold freely on the market. For further discussion see M.J. Radin, Market-Inalienability. Harvard Law Review 1987; 100: 1849.

[30] J.D. Mahoney, The Market For Human Tissue, Virginia Law Review 2000; 86: 163.

[31] At least in the Anglo-American legal tradition, property is actually treated as a "bundle of sticks," a package of rights possessed by persons relative to particular objects, including: the right to possess one's property, the right to use it, the right to exclude others, the right to transfer ownership by gift or sale, the rights to dispose of one's property after death, and the right not to have one's property expropriated by the government without payment of compensation. The legal concept of property permits fragmentation: the bundle of sticks may be separated and transferred even if the owner does not hold all of the sticks in the bundle. See, e.g., B.A. Ackerman. 1978. Private Property and the Constitution. Yale University Press, (distinguishing between the lay man's idea of property as an object and arguing that property should not be understood as a thing but as a "set of legal relations between persons governing the use of things"). See also, R. Rao, Property, Privacy, and the Human Body. Boston University Law Review 2000; 80: 359.

[32] Cf. UK Biobank, where UK Biobank will be the legal owner of the database and the sample collection, where "participants will not have property rights in the samples," but in which "UK Biobank will serve as the 'steward' of the resource, maintaining and building it for the public good." See UK Biobank, Frequently Asked Questions, at <http://www.ukbiobank.ac.uk/about/faqs.php#anonymous>. It is unclear whether under the UK Biobank policies, donors will retain any equitable interest in their samples, which may make withdrawal of samples contingent on consent of UK Biobank. Under the Biotrust Model, the right to withdraw samples would be a retained by the donor, as explained in more detail below.

[33] In American law, bodies and body parts are treated under regimes of both privacy and property, depending on the bodily material at issue and the social context. Pieces of the body like organs that are

more important for core bodily functioning, tend to be less alienable than pieces that can be detached without harm or serious consequence, such as sperm, blood, eggs, hair, etc. See Rao, op. cit. note 32. The types of materials needed to create genomic biobanks are more like the latter type of materials, for they consist mostly of blood and surgical biopsies.

[34] Boggio, op. cit. note 4, p. 46.

[35] Id.

[36] J.W. Singer. 2000. Entitlement: The Paradoxes of Property. Yale University Press: 29.

[37] In this sense, the donated property interest is also akin to a voluntarily donated easement on a piece of land, an easement being "an interest in land owned by another person, consisting in the right to use or control the land . . . for a specific limited purpose." Black's Law Dictionary, 7th Edition.

[38] J.D. Mahoney, The Market For Human Tissue, Virginia Law Review 2000; 86: 163.

[39] At least under a labor theory of value found in Locke and others.

[40] Winickoff & Winickoff, op. cit. note 3.

[41] F.W. Maitland. 1936. Equity, p. 23 (stating that the trust "is an 'institute' of great elasticity and generality; as elastic, as general a contract."

[42] Such a process of public consultation has precedent in previous public encounters with biotechnology. See, e.g., GM Nation, the UK's attempt to create a series of public consultations on genetically modified organisms. <http://www.gmnation.org.uk/>. See also admirable efforts underway as part of UK Biobank, <http://www.wellcome.ac.uk/doc_WTD003284.html>. It remains to be seen, however, how successful these particular public consultation initiatives have been or will be.

[43] E.g., in the United States, the recently enacted HIPAA Privacy Rule, 45 C.F.R. §164.501-508.

[44] This is the procedure used in many existing population studies, e.g., genetics research involving the Utah Population Database. See J.E. Wylie, G.P. Mineau, Biomedical databases: protecting privacy and promoting
research. Trends in Biotechnology 2003; 21,3:113-6.

[45] Boggio, op. cit. note 4, p. 46.

[46] Winickoff and Winickoff, op. cit. note 3.

[47] Greely would allow signed "permission" for unforeseen research with provisions stipulating the conditions of recontact, an absolute right of withdrawal at any time, time limits, limitations on availability of information to third parties, group permission requirements on top of individual permission, disclosure of commercial interests, confidentiality stipulations, and community benefits. H.T. Greely, Breaking the Stalemate: A Prospective Regulatory Framework for Unforeseen Research Uses of Human Tissue Samples and Health Information. Wake Forest Law Review 1999; 34:752–58

[48] Boggio, op. cit. note 4, p. 46.

[49] Some courts in the U.S. have recently allowed settlors to enforce the trust instrument even if the trust is not revocable. In many cases the settlor does not have standing to sue a charity for the violation of the terms of the charitable gift. See *Carl J. Herzog Foundation, Inc. v. University of Bridgeport* and G. Manne, Agency Costs and the Oversight of Charitable Organizations. Wisconsin Law Review 1999; 1999: 241-242.

[50] Some disease advocacy groups have founded their own biobanks to attract researchers to study their disease. See D. Winickoff, op. cit. note 1.

[51] Boggio, op. cit. note 4, p. 48.

[52] This term refers to Jürgen Habermas' idealized notion of a "public sphere" – a crucial political space between the state and private spheres -- in which individuals can collectively engage in deliberation, communication, identity formation, and the negotiation of norms. See, e.g., J. Habermas. 1996. Between Facts and Norms: A Discourse Theory of Law and Democracy. William Rehg, trans.. MIT Press: 298-361.

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Beyond genetic discrimination. Problems and perspectives of a contested notion

THOMAS LEMKE

Abstract

In the recent past a number of empirical studies provided evidence that increasing genetic knowledge leads to new forms of exclusion, disadvantage and stigmatisation. As a consequence, many states have inaugurated special legislation to fight "genetic discrimination".

This article focuses on some theoretical, normative and practical problems in the scientific and political debate on genetic discrimination. It puts forward the thesis that the existing antidiscrimination approach is based on the implicit idea that genes are the essence of (human) life. Since genes are held responsible for individual development and personal identity, genetic discrimination is granted a privileged legal status in comparison to other forms of discrimination. As a result the analytical and political concentration on processes of genetic discrimination may reinforce the "geneticization" of body, illness and deviance.

Beyond genetic discrimination. Problems and perspectives of a contested notion

The sequencing of the human genome at the beginning of the new century marked a symbolic milestone in the progress of genetics.[1] In the "post-genomic" era genetic research is about to transform concepts of health, illness and the body, and the practices of medicine and public health. Genetic tests already have been developed to identify the presence of particular alleles or polymorphisms that are linked to certain diseases. As the range and accuracy of these tests increase, many scholars anticipate a future when anyone may obtain genetic profile that can identify conditions for which he or she may be at elevated risk. Such knowledge should enable an individual to take preventive steps such as medication, medical monitoring, prophylactic surgery, or behavioural and environmental modification that may be wholly or partially effective to eliminating the onset of diseases.[2]

As with many other new technologies, the implementation of genetic technology raises a number of social problems. One area of concern is "genetic discrimination". As a series of empirical studies in different countries have shown the use of genetic information may lead to new forms of exclusion, disadvantaging and stigmatisation.[3] The spectrum of genetic discrimination ranges from disadvantages in work life via problems with insurance policies through to difficulties with adoption agencies. In some cases a person was turned down for a job because there were signs of a possible later illness. Likewise, health and life insurers terminated contracts or refused to conclude these if their (potential) clients were suspected of bearing the risk of a congenital disease. In other cases, couples were not allowed to adopt children if one of the two had a predisposition for a genetic illness. Experiences of genetic discrimination were also reported from healthcare agencies, the education sector and the military.[4]

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The empirical studies on the problem of genetic discrimination have not gone unnoticed. In the scholarly debate and policy discussions in society, much has been made of the danger of a "biological underclass".[5] People who were disadvantaged, pathologised and stigmatised simply owing to their genetic structure. The fear of a "new form of social prejudice"[6] led to numerous attempts to regulate the problem. Since the beginning of the 1990s a series of legislative initiatives and statements on the part of inter- and supranational organizations and commissions have been forthcoming to protect people from genetic discrimination. For example, the Council of Europe's Convention on Biomedicine (Art. 11), UNESCO's Declaration on the Human Genome (Art. 6), and the EU's Charter on Fundamental Rights (Art. 21) all explicitly prohibit discrimination on the basis of genetic features. Likewise, many nations have issued regulations designed to ensure that no one is disadvantaged on the basis of his or her genetic constitution. Thus, in some countries, including Austria and Belgium, genetic discrimination is in principle forbidden. In the United States, special laws were resolved at an early date by individual states of the Union and by the federal government. In Germany, among others, there is currently discussion on additional legal initiatives to protect against genetic discrimination.

However, it has since become ever more apparent that the concept of genetic discrimination, as is used in scholarly studies and in legal texts, entails various theoretical, normative and practical difficulties. I wish in this essay to address in greater depth four problematic areas that reveal significant gaps or weaknesses in the debate on genetic discrimination: Discrepancies in the use of the concept of discrimination in research practice (1); empirical shortcomings that mean central areas of genetic discrimination are not covered (2); conceptual problems in defining the special scientific status of genetic information (3); and finally normative ambivalences as the notion that genetic data should be subject to a more comprehensive protection compared with non-genetic information leads to unjustified unequal treatment of persons affected (4). The central argument I shall advance is that the theoretical and political-legal critique of practices of genetic discrimination frequently itself rests on the implicit notion that genes fundamentally influence human existence and form the core of our respective personalities. I suspect that this critique covertly relies on an essentialist concept of a genetic program that is deemed responsible for individual development and personal identity. In summary I shall present some conclusions drawn from this observation with regard to further scholarly and political discussion on genetic discrimination (5).

1. Definitional discrepancies: scope and content of genetic discrimination

In the scholarly literature and also in the media and the relevant laws genetic discrimination signifies the unequal treatment of people owing to actual or suspected genetic differences from the "normal" genome. Here, genetic discrimination is strictly distinguished from discrimination owing to a disability or illness.[7] All the relevant empirical studies rely on this definition of genetic discrimination, but the application of it in research practice has varied greatly – something that undermines the comparability of research findings.

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Lapham et al. use a very broad concept of discrimination for their work; it includes not only presymptomatic cases, but also cases in which the persons in question have already fallen ill.[8] As a result, phenotypical and genotypical characteristics are both used as the basis for defining the presence of genetic discrimination.[9] Most other studies seek only to speak of genetic discrimination against the persons concerned if the symptoms of the illness are very mild or do not constitute a disability or restriction in performance.[10] By contrast, an extremely narrow concept of discrimination is utilized by Mark A. Hall and Stephen S. Rich in their study, as they only take into account cases in which the persons concerned were completely presymptomatic, in other words in which the illness was in no manner manifest yet. The different concepts of discrimination lead to contrary findings: While Hall and Rich had difficulty documenting even a single case of genetic discrimination, the study by Lapham et al. states that almost half of those polled had experienced genetic discrimination.[11]

Not just the scope, but also the substance of the concept of discrimination is a bone of scholarly contention. In the relevant studies, genetic discrimination is regularly construed as the "unjustified unequal treatment of persons owing to their genetic characteristics".[12] The literature usually takes a critical perspective: The implicit assumption is that unequal treatment owing to factual or suspected genetic differences is legally impermissible and/or morally objectionable. However, in addition to this majority view, there are other opinions that consider genetic discrimination a factually legitimate form of risk differentiation.[13] An extreme position in this regard is taken in the libertarian argument advanced by Colin S. Diver and Jane Maslow Cohen. They contend that the unequal treatment of human beings owing to genetic factors is not only morally unproblematic and legally permissible, but also socially necessary as an economic instrument of resource allocation and risk calculation. From this standpoint, government prohibitions on genetic discrimination prevent the efficient market regulation that requires a "regime of genetic transparency".[14]

Needless to say, the conceptual discrepancies are not only of interest as regards research strategies and at the theoretical level, but are of great significance for legislative practice and social policy: Is genetic discrimination a legitimate means of risk differentiation in order to generate economic growth and prosperity or a social evil that calls for corresponding government measures to protect persons from it? And if the latter is the case, what individuals and groups with what illnesses and/or risks of illness should be protected against discrimination, and in what way? How broad or narrow should the circle of those be who are to benefit from legal protection?

2. Empirical deficits: "Everyday eugenics" and indirect discrimination

The studies to date that deal with the problem of genetic discrimination have a clear emphasis. They concentrate on institutional actors disadvantaging individuals and their relatives. This analytical focus means that important practical areas of genetic discrimination do not get equal attention. I shall scrutinize two empirical deficits more closely here: the focus on institutional actors and the neglect of indirect forms of discrimination.[15]

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2.1. Focus on institutional actors and asymmetric decision-making

So far studies of genetic discrimination mainly focused on institutional actors, on the one hand, and individuals and relatives, on the other. This juxtaposition is too strongly entrenched in the juridical frame of committer/victim. However correct and important it may be to expose discriminating practices by insurance companies, employers, adoption agencies and other organizations, this disregards a decisive arena of genetic discrimination: disrespect and stigmatisation by family, friends and fellow human beings. This "everyday discrimination" is systematically ignored in the studies to date, meaning a key field of genetic discrimination remains excluded.

In addition to this analytical gap there is another problem: Literature on genetic discrimination tends to concentrate on the "negative" pattern of litigation: on coercive measures and asymmetric decision-making processes. The guiding idea here relates to organizations that refuse to sign contracts or reject qualifications. Thus, the studies exclude to investigate to what extent formally voluntary options for action and symmetrical decision-making situations can have a discriminating effect. A core element of the new genetic knowledge consists precisely of the fact that it transforms natural constrains into new individual options – options that may engender new constrains. Scholars discern a transition from a compulsory eugenics to a more indirect form of control and guidance of individuals, something the academic debate has termed "voluntary"[16] "individualist"[17], "liberal"[18] or "everyday eugenics"[19]. Those critical accounts point to the fact that individual decisions and private choices may, taken collectively, have negative social effects and create new pressures on reproductive behaviour. While some scholars resist the idea that human genetics represents a disguised or a new kind of eugenics, the question remains whether the phenomenon of genetic discrimination is restricted to explicit prohibitions and rare exceptions, or may also result from "normal" risk assessments, "rational" concepts of health and the idea of a "self-regulative behavioural management".[20]

2.2. Neglect of indirect mechanisms and structural links

This conceptual problem points to the limits of a person-centred and case-oriented notion of discrimination. Alongside discrimination of people who are directly confronted by disadvantaging or stigmatisation, forms of "indirect" discrimination must also be considered. This should include all factors that indirectly influence the persons involved and constrain their scope for decision-making and their options for action. While direct genetic discrimination remains a matter of individual cases and describes the way in which certain people with genetic characteristics are treated as individuals, indirect discrimination refers to social judgments of unworthiness, structures of prejudice and forms of disrespect addressed to all members of society.[21]

A comprehensive analysis of genetic discrimination must also focus on those strategies for action with which the persons affected anticipate negative categorization by their social environment and adapt their behaviour accordingly. The most effective means of pre-empting genetic discrimination entails not drawing genetic risks to the attention of other people, let alone institutional actors such as insurance companies or employers. This "information control" runs from the choice of spouse via neighbourly relations to working life. The persons affected feel the act of excluding their own risk

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of falling ill and the related fears as a form of compulsion, as a restriction in forms of communication, and as the necessity to keep important information on themselves and their own future secret from others.[22]

Moreover, the judgment on unworthiness as is expressed in practices of genetic discrimination, also impacts on decisions on reproduction that go far beyond the realm of those who are directly affected by the risk of genetic illness. While studies on the problem of genetic discrimination to date have concentrated on postnatal genetic tests and the disadvantaging of persons already born, it bears asking whether the analysis should not also cover the field of prenatal diagnostics (selective abortion on the basis of genetic indicators) and pre-implantation diagnostics (deliberate choice of genetically "desired" embryos).[23]

But it does not suffice for a systematic analysis of genetic discrimination to simply internally differentiate between various practices of genetic discrimination. We must also consider the "structural links" between different forms of discrimination. In this way, we can study how forms of genetic discrimination link up with sexist and racist practices and mutually reinforce one another.[24]

As regards the relationship of genetic discrimination and racism, it bears stating that some genetic illnesses are encountered more frequently in certain ethnic groups or groups of the population than in others. For example, sickle cell anemia occurs more frequently among persons of African descent, Tay-Sachs Syndrome is especially widespread among Ashkenazi Jews, and most persons with the beta thalassemia gene are inhabitants of the Mediterranean rim. Since certain ethnic groups are differently susceptible to specific genetic illnesses, there is a danger that members of minorities will be associated with such genes and treated pathologically, even if they do not bear the particular genetic mutation. For example, sickle-cell anemia in the United States is considered a disease of Afro-Americans, although it is to be found just as frequently among population groups from the Mediterranean rim.[25] There is likewise the danger that there will be a disparity in resource allocation and in the public attention on these diseases. For example, in the USA research into cystic fibrosis, which affects primarily white coloured people, receives far more financial backing than does research into sickle cell anemia, even if the latter is far more widespread among the population as a whole.[26]

The practice of *gender verification* at sports competitions is a striking example of the linking of genetic discrimination und sexism. Gender control by genetic testing emerged as an issue in the 1960s when there were rumours that men had passed themselves off as women to take part in women's competitions. Despite the intensive criticism down through the years, genetic analyses are still undertaken to verify gender in sport. However, only women and not men have to have their gender verified, and suffer the possible consequences of "failing" such a test. Although almost all International Olympic Committees now forego such verification procedures, at world championships for some sports gender continues to be genetically "tested". For example, at the Volleyball World Championship in 2002 in Germany, all the women were subjected to a genetic analysis if they did not have test results from a prior sports competition.[27]

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A person-centred and case-oriented concept of discrimination is not able to explore the systematic links between racist and sexist practices and ideologies on the one hand, and patterns of geneticist interpretation and action, on the other. Genetic discrimination does not entail isolated and chance deviations from the norm, individual cases and erroneous institutional developments, but it points to social practices that divide persons into genetic categories and promote a belief in the determining power of genes.[28] Unfortunately notions of genetic essentialism frequently also shape the analysis and critique of genetic discrimination, leading to conceptual confusion on the one hand and normative ambivalences on the other.

3. Conceptual confusions: the exceptional epistemological status of genetic information

Studies on genetic discrimination and legislative initiatives to protect affected persons share a common presumption: both assume an exceptional status of genetic information in two ways.[29] First, there is the claim that genetic information differs epistemologically from non-genetic information. From this perspective there is a clear scientific line dividing genetic testing from non-genetic procedures, genetic diseases from non-genetic conditions. Second, there is the suggestion that genetic information should be normatively distinguished from non-genetic information: discrimination on the basis of genetic data on the current or future health of a person is then unjust or more unjust compared to discrimination on the basis of non-genetic medical information. As I shall attempt to show, both assumptions are untenable. I shall focus in this section on the justifications for an exceptional epistemological status of genetic information, and then in the next section trace the normative problems to which this gives rise.

3.1. Genetic exceptionalism

There are at least three arguments put forward to justify the exceptional (medical) position of genetic screening. Genetic analytical procedures are claimed to be more precise than other medical tests as they provide predictive information on the health of an individual. They are said to allow it to diagnose with certainty or greater probability whether a person will fall ill with a specific disease. Second, the results of genetic tests are said to enable conclusions to be drawn on the state of health or risk of illness of relatives of the person screened. Genetic tests are, thirdly, believed to differ from traditional diagnostic techniques and conventional medical methodologies, as they purportedly reveal fundamental personal characteristics of the person examined. All three criteria thus used to justify “genetic exceptionalism” do not stand up to closer scrutiny.[30]

First, only a very few genetic tests allow predictive statements to be made on future illnesses. Genetic illnesses are as a rule characterized by incomplete penetration and/or variable expressivity.[31] The former relates to the frequency with which genetic mutation actually trigger the corresponding disease in the person bearing the gene. In the case of complex illnesses such as cancer, Alzheimer’s or diabetes the presence of a changed gene does not necessarily lead to illness. For example, proof of a mutation of one of the so-called BRCA genes increases the statistical probability of a woman developing breast cancer, but the question whether, when and in what way she

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succumbs to the illness is by no means answered.[32] As recent studies have shown, even monogenetic illnesses such as cystic fibrosis or Morbus Huntington do not entail 100% penetration.[33]

Variable expressivity refers to the fact that the symptoms of one and the same illness can differ vastly from one individual to the next, although both possess the same mutated gene. In other words, the same DNA mutation can trigger quite different clinical symptoms – or none at all; conversely, one and the same disease can be triggered by different genetic variations.[34] It bears stating here not only that the predictive value of genetic information is regularly exaggerated, but also that non-genetic medical tests provide information on future health risks, such as a HIV test, a cholesterol screen, or the proof of an asymptomatic hepatitis B infection. Likewise, blood-pressure tests or proof of blood in stool are of great diagnostic importance when identifying illnesses at an early stage before the first symptoms of coronary heart disease or stomach cancer have arisen.[35]

As regards the second criterion, it is also difficult to recognize the medical uniqueness or even the special status of genetic analyses. As Murray rightly emphasizes, people have long since known that their own risk of having an illness increases if the same illness has already arisen in their family. Likewise, in the past, doctors have relied on information on relatives to supplement their medical knowledge on a particular patient. Moreover, a person's family history has for some time been factored into insurance contracts and/or consulted when defining premiums. Genetic tests are, in other words, neither the only nor the most important tools for using medical data on relatives in order to reach statements on an individual's health status.[36]

The third criterion differentiates between a fateful and immutable genetic predisposition, on the one hand, and chance and controllable environmental factors, on the other. This argument rests on the notion that genes are autonomous and active, forming a kind of control centre that steers and regulates the organism.[37] This assumption is at best a vast simplification if not misleading or false. First, genes are not static units, but an integral part of a complicated biochemical network that is defined by the dynamic interaction of interdependent actors.[38] For this reason, it is an impermissible simplification if we assume that a characteristic or function is determined in part by heredity and otherwise by environmental influences. This fails to consider that genetic changes can also be first *acquired* in the course of life owing to environmental influences and a specific life style.[39] Furthermore, there can be no simple equation between genetic causality and an inevitable fate and/or non-genetic factors and personal scope to take free decisions. Neither do genetic causes of illness signal a necessary biological fate, nor are non-genetic conditions in principle easier to control than their genetic counterparts or the result of personal choices for which the individual is responsible.[40] On the contrary, there are also many non-genetic factors which we as individuals can by no means control: "If the air we breathe and the water we drink are polluted, if our parents or co-workers are heavy smokers, if we are reasonably prudent but injured in an accident nonetheless, it is hard to say that we bear any significant measure of responsibility for the resulting illness".[41]

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All the three criteria that ostensibly justify the special (medical) position of genetic information discussed here thus prove unconvincing. That said, not only is the line dividing genetic from non-genetic factors is diffuse, but also it is not clear what exactly "genetic" means. Put differently: What we have here is thus not a definitional problem that results from an impermissible conflation or erroneous shift of the line dividing two essentially distinguishable components (genetic/non-genetic); instead, it is a systematic problem that lies in the "nature" of the matter at hand. What is respectively termed a "gene" or "genetic" entails great semantic flexibility and depends on the respective scientific definition and social context.[42]

3.2. Genetic or non-genetic?

The concept of genetic illness has been constantly expanded in recent decades. Today, genetic factors are considered to be responsible not only for "monogenetic" illnesses (so-called congenital illnesses), but also for multifactored diseases such as cancer, Alzheimer's, diabetes and many other widely-spread illnesses. This semantic expansion has gone hand in hand with a shift in meaning such that illnesses are increasingly construed as genetic deviations from the norm. However: If every illness can potentially be ascribed to genetic changes, it becomes questionable what exactly the contribution made by genetic factors is and/or how genetic causes of illnesses are to be distinguished from their non-genetic counterparts.[43]

It is not only impossible to finally and definitively state what distinguishes a genetic illness from a non-genetic complaint; it is likewise unclear how genetic analyses can be distinguished sharply from non-genetic analyses. "Genetic diagnostics" is the umbrella term used to cover all types of diagnosis of genetically determined illnesses and features. This includes tests at the level of DNA as well as phenotypical diagnoses (such as the test for genetically conditioned colour blindness), chromosome examinations and analyses at the level of genetic products.[44]

Joseph S. Alper and Jon Beckwith rightly point out that this comprehensive definition of genetic tests applies to practically all clinical test procedures. They emphasize that most medical tests focus on diagnosing abnormal concentrations of biochemical units. Typically, these units are proteins that are generated directly by genes, or molecules whose synthesis depends to a more or less large degree on the activity of the genes. Which is why a striking biochemical finding can be the result of a changed gene or genotype. Even if these non-genetic tests focus on determining the function of the various organs, they possibly also provide information on genetic characteristics. In addition, in some cases only the medical context can decide whether a genetic or a non-genetic test is involved: "A test of blood cholesterol concentration may be regarded as genetic when testing an individual with a family history of hypercholesterolemia, a single gene recessive disorder. The same test is clearly a non-genetic medical test when ordered in the course of a routine physical examination."[45]

It follows that genetic diseases cannot be exclusively detected by means of genetic testing (or, to be more precise, DNA diagnostics). In many cases, conventional screening procedures can reliably prove the existence of a genetic condition. The fact that a distinction between genetic and other diagnostic devices is not merely a technical matter can be seen from the history of the PKU screening as reconstructed

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by Diane Paul. PKU is a metabolic illness where an enzyme defect prevents the amino-acid phenylalanine being transformed into tyrosine. However, initiating a special diet when the patient is still a child can largely offset this problem. When, in the 1960s, the so-called Guthrie Test was developed and used as a diagnostic tool with newborn infants, the genetic dimension of the disease played no role; instead, back then PKU was considered a treatable form of mental retardation. Moreover, the method used to prove its existence is a biochemical test, not DNA-level screening. Only much later did the Guthrie Test come to be regarded as a genetic test and PKU as a genetic condition – a label that served various interests at the same time. The molecular medicine visionaries were able to point to the PKU Test to demonstrate that genetic tests exist that does provide a therapeutic benefit. That said, critics of genetic screening also profited from the label "genetic test". In the framework of the Human Genome Project, commissions were set up in the United States and various other countries to specifically regulate genetic analyses (in contrast to other medical tests). Since genetic screening requires greater technical care and more intensive legal regulation than do other methods, the definition of the PKU Test as a genetic test was also of interest to critics of genetic determinism who are sceptical of the increasing use of genetic screening.[46]

It is obviously not only difficult to distinguish with any scientific precision between genetic and non-genetic diseases. Moreover, the dividing line between genetic and non-genetic tests can also not be defined intrinsically in technical terms. As we saw, in the final instance the difference is the product of social negotiations and scientific compromises. This insight has a strong impact on political initiatives that endeavour to regulate the use of genetic data by means of special anti-discrimination legislation.

4. Normative ambivalences: the legal privilege of genetic information

Discrimination on the basis of genetic factors is directed against "asymptomatic ill persons"[47], while discrimination of ill and disabled persons results in unequal treatment on the basis of phenotypic features. Generally, the former is considered more problematic in moral and legal terms than the latter, which leads to a asymmetrical treatment of those suffering from discriminatory practices. First, genetic and non-genetic discrimination are treated differently in legal terms, which prompts the question what criteria are used to justify such an unequal treatment of persons who are equally affected by discriminatory practices. Second, there is the danger that the exceptional legal status of genetic discrimination simply "normalizes" all non-genetic forms of discrimination. If in the widest variety of different social areas the disabled and the ill are regularly discriminated against compared with the healthy, then this appears legitimate to the extent special protection exists for persons who are affected by practices of genetic discrimination.[48] Put differently: the concentration on the legal impermissibility and moral reprehensibility of genetic discrimination isolates different forms of discrimination, plays them off against one another, and threatens to augment the social acceptance of practices of non-genetic discrimination.

Gregor Wolbring has pointed out that in principle two completely different strategies for legal policy are conceivable in order to prevent discrimination against asymptomatic ill persons. The one path could be to extend and expand existing anti-discrimination legislation on the equal treatment of the disabled such that it equally

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covers discrimination against the asymptomatic and symptomatic, the genetic and the non-genetic ill. However, at present a quite different approach is being taken in the United States and many other countries that envisage special laws to protect the asymptomatic ill. The laws or legislative projects currently being negotiated are based on a strict and unequivocal distinction between the asymptomatic and symptomatic ill. Wolbring points out that this serves to increase the legal gap between the two groups of persons affected. Instead of grasping genetic discrimination as an integral part of a social continuum of discriminatory practices, it is considered as a specific caesura to be distinguished in conceptual and normative terms from other forms of disadvantaging. As Wolbring rightly remarks, it is however questionable whether discrimination against the asymptomatic ill would exist without discrimination against the symptomatic ill and the disabled.[49]

Strikingly, there are differences in the legal and moral appraisal not only as regards the symptomatic as opposed to asymptomatic ill, but also within the group of the asymptomatic ill. A distinction is made there between persons affected by genetic risks and those exposed to non-genetic risks of illness. Both sections of the group share the fact that the illness is not yet developed, and will possibly never do so, but the legal evaluation of the risks is completely different. The problem is perhaps best illustrated by juxtaposing two similar cases that were decided almost simultaneously in the same federal state of Germany.

4.1. "From the cradle" – The rebirth of genetic essentialism as anti discrimination policy

The first case was recently taken up by various media and found a lot of attention inside and outside Germany.[50] In August 2003, the State of Hessen refused to employ a teacher as a civil servant after she had completed her trial period. The enquiry by the officially appointed occupational physician had revealed that the young woman's father suffered from Huntington's Disease.[51] The report came to the conclusion that at the present point in time the applicant's health was suitable to enable her to take up the job, but she was barred from becoming a civil servant on the ground that there was an increased probability that she would fall ill in the foreseeable future and become enduringly unfit to discharge her duties. The applicant lodged an appeal against this decision before the Administrative Court in Darmstadt, which ruled mainly in her favour and instructed the State of Hessen to immediately appoint her to government service. In the court's opinion, the school authorities had wrongly assessed the state of her health as an applicant, as they had claimed that the 50-percent risk of illness meant there was a "most strong probability" she would enduringly not be able to discharge her duties. The school board declined to contest the decision and has since employed the woman in question under a government service contract.[52]

At the same time, another case was before the courts but attracted far less public attention. Again in Hessen, a young man was dismissed while still on probation for government service, as in the opinion of his employers given his weight of 120 kg his health was not suited for a career in general administration. Here again the applicant took the matter before the courts. However, in its ruling the Frankfurt Administrative Court confirmed that the dismissal was legal as the employer was permitted to pre-empt the risk of having to foot the bill for later enduring damage to the man's health.[53]

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In other words, although the applicant was not yet ill and it is completely uncertain whether and in what way his elevated body weight might impair his health in the future he was treated in legal terms as though he were already incapable of pursuing his duties.

These examples show that the distinction between genetic and non-genetic information cannot serve as the basis for legal differentiation. First, it is not intelligible why, for example, the use of biochemical methods that allow making inferences on a person's genetic disposition is permissible for discriminatory purposes, while DNA tests, that reach the same results, are forbidden. It would seem not only fairly impractical but also unfair to prohibit an insurance company from evaluating a genetic analysis for a complex disease, while the results of a non-genetic test for the same illness may be relied on. This methodology essentially creates a legal situation in which people with positive genetic diagnoses receive more protection against discrimination and data abuse than those whose findings are based on non-genetic methods: "Would the law mean that the records of a person with a presymptomatic heart condition who was given a genetic test for some mutant gene associated with heart disease would be covered by anti-discrimination provisions, but not the records of a person with the same condition whose physician order only nongenetic tests?"[54]

Second, it remains unclear why institutional actors such as insurance companies or employers should be forbidden from using *a* source of genetic information (genetic screening) although they are allowed to draw on other forms of genetic knowledge. For example, according to a draft by the German Health Ministry for a bill on genetic diagnostics, an insurance company would be prohibited from demanding that a woman who a BRCA test shows as positive for breast cancer pay a higher insurance premium, whereas this is permissible with regard to a woman who has preferred not to undergo genetic screening but in whose family several women have already had breast cancer.[55] This asymmetric decision-making principle not only violates the principle of fairness, but also means we must fear that such a regulation will compel people to opt for genetic analyses even if they did not originally want to – for example, in order to get insurance policies (at standard conditions).[56]

Third, the increasing discovery of genetic factors for the genesis of illnesses will in future make it ever harder to draw a line between genetic and non-genetic conditions. With reference to the above-mentioned case, a series of research findings could be cited, for example, that point to a genetic component in obesity.[57] What would the judgment be if an applicant could credibly claim that a specific genetic disposition is (co-) responsible for his increased body weight? Would we then be confronted by a case of genetic discrimination and would need to specially protect those concerned from it?

To summarize, we can say that the emphasis of anti-discrimination policies is evidently more on the (genetic) "nature" of information and less on social practices in which these data are used and evaluated.[58] The legislation to protect against genetic discrimination are not least the product of the implicit notion that genes fundamentally influence human existence and constitute the core of each personality. It is accordingly unfair to punish persons for something that they cannot themselves

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control.[59] This logic is also to be found in the justifications given by the German Health Ministry for the planned legislation on genetic diagnostics. The statement in question says that any form of genetic discrimination must be countered: "For our genetic characteristics 'are with us in the cradle' and we are therefore not responsible for them".[60] This argument may initially seem plausible, but on closer inspection is not very convincing, as it is based on the idea of a genetic program that is "responsible" for the development and identity of individuals and that both constitutes and constraints their scope for action. It is the assumption of a particular power and autonomy of genetic factors that forms the basis for their privileged legal status. In principle, there are many non-genetic factors that are just as little susceptible to personal control although we do not call for them to be specifically protected. It is by no means evident why a person whose higher risk of contracting a specific type of cancer, say, is attributable to genetic factors, should enjoy greater protection than some one whose health is threatened by environmental factors such as poor working conditions or polluted air. Should not this group of persons likewise be effectively protected?[61]

We can assume that the explicit withdrawal of responsibility in the case of genetic risks of illness is the other side of the coin of an increasing ascription of responsibility for all non-genetic factors.[62] The reductionist concept of genetic fatalism that purportedly unravel automatically and independently of the individual person's will contrasts with the radicalized appeal to personal responsibility and personal accountability as regards health and the prevention of illness.[63] Possibly, the different judgments in the two afore-mentioned cases stem from a shared underlying logic that places the question of personal responsibility at the centre of things. Increased body weight and the resulting risks to a person's health are essentially considered the (erroneous) result of individual choice, while the risk of contracting Morbus Huntington is viewed as biological fate and thus treated as something for which a person cannot be held responsible.

5. Critical paradoxes

The call to distinguish genetic discrimination from other types of discrimination and subject it to special legislation has a paradoxical impact. The prohibition on the "unequal treatment" of people with an "abnormal" genetic constitution reinforces the cultural belief in the exceptional status of genetic factors, something which the legal regulation was supposed to counter in the first place. The analysis and critique of genetic discrimination itself relies on the phantasm that genes forms the "blueprint" for an individual and the "secret of life".[64] In this way, the anti-discrimination legislation threatens to intensify the very problem it set out to solve. This brings us up against a key dilemma.[65] On the one side, there are practices of genetic discrimination and people who suffer from these practices and, on the other, genetic essentialism is rejuvenated and reinforced by the academic and legal confirmation of the special role of genetic factors.

This dilemma by no means signifies that legal stipulations to protect people with genetic peculiarities are superfluous or even damaging. On the contrary, even if genetic information, seen scientifically, should be accorded no privileged role over non-genetic data, in social reality they quite obviously have a pronounced

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importance.[66] In cultural terms, genes symbolize something fateful and immutable[67], are considered "the most intimate biological property that we have"[68] and are thought to decisively influence the individual course of life[69]. Unlike other health risks, that are temporary, treatable, and can essentially be eliminated, genes are construed as the basis for a person's identity. He or she is said to "carry" or "possess" not only genetic risks, these are actually considered to be an integral part of the person's physical existence.[70] If genetic factors are made responsible for a disease, then the person affected feels they are not controllable and are thus more threatening than if non-genetic reasons are cited.[71] Equally, the risk of genetic illness cannot be separated from the history of eugenics, the cataloguing and murdering of people who were considered "genetically inferior", from a trans-generational notion of illness and the idea of "defective" or "poor" genes.[72] As long as this cultural stereotype and historical prejudice persists, and people are disadvantaged or shown disrespect owing to their genetic peculiarities, it is imperative to legally protect them.

However, such legal protection must not result in genetic data being isolated form other (predictive) medical information. Genetic discrimination is the result of an increasing extension of the concept of illness and disability and the expansion of existing practices of contempt, stigmatization and exclusion. For this reason, the prohibition on genetic discrimination must necessarily be supplemented by profound institutional reforms and comprehensive regulations that more effectively protect persons already ill or disabled from social exclusion and disadvantage.[73] That said, it is also necessary to rethink the analysis and critique of genetic discrimination with a view to its premises and goals. Otherwise we would run the risk of the distinction between genetic and non-genetic leading us to ignore the more fundamental question of the way (predictive) medical information is in general used to categorize persons, attribute characteristics and features to them, and exclude them from particular benefits.[74] In the absence of such dual self-enlightenment, the critique of genetic discrimination simply leads to a further "geneticization" of the body, illness and deviance.

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[2] See e.g. F. S. Collins, Shattuck Lecture – Medical and Societal Consequences of the Human Genome Project, in: New England Journal of Medicine. 1999; 341: 28-37.

[3] P. R. Billings, M. A. Kohn, M. de Cuevas, J. Beckwith, J. S. Alper & M. R. Natowicz, Discrimination as a Consequence of Genetic Testing, in: American Journal of Human Genetics. 1992; 50: 476-482; L. N. Geller et al., Individual, Family, and Societal Dimensions of Genetic Discrimination: A Case Study Analysis, in: Science and Engineering Ethics. 1996; 2: 71-88; L. Low, S. Kind, T. Wilkie, Genetic discrimination in life insurance: empirical evidence from a cross sectional survey of genetic support groups in the United Kingdom, in: British Medical Journal. 1998; 317: 1632-1635; M. Otlowski, S. Taylor, K. K. Barlow-Stewart, Australian Empirical Study into Genetic Discrimination, in: Eubios. Journal of Asian and International Bioethics. 2002; 12: 164-167.

[4] For an overview of the studies available to date on experience with genetic discrimination, see T. Lemke/C. Lohkamp, Formen und Felder genetischer Diskriminierung. Ein Überblick über empirische Studien und aktuelle Fälle, in: Wolfgang van den Daele (ed.), Biopolitik, Wiesbaden: Verlag für Sozialwissenschaften; 2005: 45-70.

[5] D. Nelkin, L. Tancredi. Dangerous Diagnostics. The Social Power of Biological Information, Chicago/London: University of Chicago Press; 1994, p. 176.

[6] J. Rifkin, Genetische Diskriminierung. Eine neue Form des sozialen Vorurteils, in: Süddeutsche Zeitung, 29th june 2000.

[7] "[...] genetic discrimination is defined as discrimination against an individual or against members of that individual's family solely because of real or perceived differences from the 'normal' genome of that individual. Genetic discrimination is distinguished from discrimination based on disabilities caused by altered genes by excluding, from the former category, those instances of discrimination against an individual who at the time of the discriminatory act was affected by the genetic disease" (Billings et al. op. cit. note 3, p. 477; see also M. R. Natowicz et al. Genetic Discrimination and the Law, in: American Journal of Human Genetics. 1992; 50: 465-475, here: p. 466).

[8] V. E. Lapham, C. Kozma, J. O. Weiss. Genetic Discrimination: Perspectives of Consumers, in: Science. 1996; 274: 621-624.

[9] See also the comprehensive definition proposed by T. Neuer-Miebach. "Genetische Diskriminierung". In: CDU-Bundesgeschäftsstelle (ed.), Arbeitsmaterialien Bioethik, Berlin; 2001: 53-67, here: p. 54: "I term disadvantaging on the basis of a genetic predisposition that leads to disability and/or illness, or a genetically caused illness, a risk of disability or illness, genetic discrimination."

[10] Billings et al. op. cit. note 3; Geller et al. op. cit. note 3.

[11] M. A. Hall, S. S. Rich. Laws Restricting Health Insurers' Use of Genetic Information: Impact on Genetic Discrimination, in: American Journal of Human Genetics. 2000; 66: 293-307; here: p. 294; D. Hellman. What Makes Genetic Discrimination Exceptional? in: American Journal of Law & Medicine. 2003; 29: 77-116, here: p. 86.
William Nowlan and Philip R. Reilly also champion a narrow interpretation of genetic discrimination and consider the problem's importance to be vastly exaggerated (W. J. Nowlan. A Scarlet Letter or a Red Herring? Genetic Discrimination Is of Little Concern Compared With Existing US Healthcare Problems, in: Nature. 2003; 421 (6921): 313; P. R. Reilly. Genetic Discrimination, in: Clarissa Long (ed.), Genetic Testing and the Use of Information, Washington DC: AEI Press; 1999: 106-132.

[12] Deutscher Bundestag. Schlussbericht der Enquete-Kommission "Recht und Ethik in der modernen Medizin", Opladen: Leske und Budrich; 2002, p. 288.

[13] For a brief juxtaposition of the two poles in the discussion, see A. Somek. Genetic Discrimination, in: Society. 2003; 40 (6): 35-43; here: p. 37-39.

[14] C. S. Diver, J. M. Cohen. Genophobia: What Is Wrong With Genetic Discrimination? in: University of Pennsylvania Law Review. 2001; 149: 1439-1482.
For example, the authors hold that in the case of employment relationships the use of genetic investigative techniques could lead to employees who are unproductive or prone to illness not being employed in the first place and thus to costs being saved: "Hiring or promoting an under-qualified or under-productive worker is inevitably costly to the employer. [...] Monitoring and corrective action require investment in supervision, and often require changes in production design or scheduling. In the meantime, the under-performing worker inflicts on the organization both demoralization costs and the opportunity costs of foregone output. For these reasons, it is almost always in the employer's interest to establish better ex ante screening mechanisms so as to select workers who will require less supervision and corrective action." (Diver/Cohen op. cit., p. 1461).

[15] The following considerations are based primarily on the results of the empirical study I undertook on experiences people affected by Huntington's Disease have had of genetic discrimination (T. Lemke. Genetische Diskriminierung in Deutschland – Eine explorative Studie am Beispiel der Huntington-Krankheit, in: Soziale Welt. 2005; 56: 417-440).

[16] L. Wess. Eugenik im Zeitalter der Gentechnologie – Vom Zwang zur freiwilligen Inanspruchnahme, in: Anne-Dore Stein (ed.), Lebensqualität statt Qualitätskontrolle menschlichen Lebens, Berlin: Marhold; 1992: 65-82.

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[17] A. Waldschmidt. Das Subjekt in der Humangenetik. Expertendiskurse zu Programmatik und Konzeption der genetischen Beratung 1945-1990, Münster: Verlag Westfälisches Dampfboot; 1996: 275.
[18] J. Habermas. Die Zukunft der menschlichen Natur. Auf dem Weg zu einer liberalen Eugenik? Frankfurt am Main: Suhrkamp; 2001.
[19] T. Degener, S. Köbsell. "Hauptsache, es ist gesund"? Weibliche Selbstbestimmung unter humangenetischer Kontrolle, Hamburg: Konkret Verlag; 1992: 67-92.
[20] P. Weingart. Politik und Vererbung: Von der Eugenik zur modernen Humangenetik, in: Eckart Voland (ed.), Fortpflanzung: Natur und Kultur im Wechselspiel. Versuch eines Dialogs zwischen Biologen und Sozialwissenschaftlern, Frankfurt am Main: Suhrkamp; 1992: 28-50; here: p. 45-49.
[21] See the distinction made by the German Government Enquiry Commission on "Right and Ethics in Modern Medicine": "Direct discrimination means a morally not justified unequal treatment or ostracization of persons by other persons or institutions. This covers, for example, discrimination of employees or insured persons or disabled persons on the basis of a genetic test. Indirect discrimination means the social values and norms that express a disrespect for certain people. This would include the establishment of social norms such as 'a life worth living' owing to chronic illness or disability" (Deutscher Bundestag. op. cit. note 12, p. 57).
Deborah Hellman's "expressivist argument" puts forward a similar distinction (op. cit. note 11, p. 108).
[22] Cf. M. Konrad. From Secrets of Life to the Life of Secrets: Tracing Genetic Knowledge as Genealogical Ethics in Biomedical Britain, in: Royal Anthropological Institute. 2003; 9: 339-358.
[23] T. Neuer-Miebach, op. cit. note 9, p. 56-59; S. Volz. Diskriminierung von Menschen mit Behinderung im Kontext von Präimplantations- und Pränataldiagnostik, in: S. Graumann, K. Grüber (eds.), Medizin, Ethik und Behinderung, Frankfurt am Main: Mabuse Verlag; 2003: 72-88.
[24] S. M. Wolf. Beyond 'genetic discrimination': Toward the broader harm of geneticism, in: Journal of Law Medicine & Ethics. 1995; 23: 345-353; L. B. Andrews. Future Perfect. Confronting Decisions About Genetics, New York: Columbia University Press; 2001: 77-97.
[25] T. Duster. Backdoor to Eugenics, New York/London: Routledge; 1991: 24-28; 45-51.
[26] European Commission (ed.). Ethical, legal and social aspects of genetic testing: research, development and clinical applications, Brussels: EC; 2004: 50.
On the increasing focus on the category of "race" in genome research see H. Bradby. Genetics and racism, in: T. Marteau, M. Richards (eds.), The troubled helix: social and psychological implications of the new human genetics, Cambridge: Cambridge UP; 1996: 295-316; G. T. Ellison, T. H. George, I. Rees. Social identities and the 'new genetics': scientific and social consequences, in: Critical Public Health. 2002; 12: 265-282; P. Aldhous. Geneticist fears "race-neutral" studies will fail ethnic groups, in: Nature. 2002; 418: 355-356; J. S. Alper, J. Beckwith. Genetics, Race, and Ethnicity, in: J. S. Alper et al. (eds.), The Double-Edged Helix. Social Implications of Genetics in a Diverse Society, Baltimore/London: John Hopkins UP; 2002: 175-196; Nature Genetics 2004: 'Race' and the human genome, in: Nature Genetics Supplement 36 (november 2004).
The danger of a linkage of genetic discrimination and racism is obviously not restricted to the clinical domain. Racist discrimination can arise from the use of genetic data in criminal justice, something the inventor of the "genetic finger-print", Sir Alec Jeffreys, points to. Jeffreys expressed concern given the ever more extensive DNA database run by the Metropolitan Police Force that now holds 2.5 million data sets. They include not only DNA samples from convicts, but also genetic information on suspects. "For a start, we are now putting not just criminals but suspects in our database, and this is clearly very highly discriminatory. If you go to certain places such as South London, you will get suspects who are predominantly black. Similarly you will get a lot of Asian suspects in Birmingham." Jeffreys concludes that the national database will be filled with a large number of blacks and persons of Asian descent who have not been found guilty of a crime, but of whom one suspects that they will in the future commit such (cf. R. McKie. Inventor warns over abuse of DNA data, in: Observer, 8th august 2004; see also S. Cole. Suspect Identities. A History of Fingerprinting and Criminal Identification, Cambridge, MA & London: Harvard University Press; 2001: 287-311; D. Wasserman, R. Wachbroit. Genetics and Criminal Behavior, Cambridge: Cambridge University Press; 2001.

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[27] See the statement of Albert Fromme, Director of the Medical Section of the Organization Committee in Münster handling the Volleyball World Championships: "For the sportswomen, this is a form of discrimination. Men do not have to prove their masculinity" (quoted from Sport 1. Fromme: "Männer müssen ihre Männlichkeit nicht beweisen", in: www.sport1.de/coremedia/generator/ www.sport1.de/ Sportarten/Mehr/Volleyball (access date: 4th september 2002).
See also J. L. Simpson, A. Ljungqvist, M. A. Ferguson-Smifh, A. de la Chapelle, L. J. Elsas, A. A. Ehrhardt, M. Genel, E.A. Ferris, A. Carlson. Gender Verification in the Olympics., in: Jama – Journal of the American Medical Association. 2000; 284 (12): 1568-1569; B. D. Dickinson, M. Genel, C. B. Robinowitz, P. L. Turner, G. L. Woods. Gender Verification of Female Olympic Athletes. Medicine and Science, in: Sports and Exercise. 2002; 34: 1539-1542.
[28] Wolf, op. cit. note 24.
[29] J. S. Alper, J. Beckwith. Distinguishing Genetic from Nongenetic Medical Tests: Some Implications for Antidiscrimination Legislation, in: Science and Engineering Ethics. 1998; 4: 141-150.
[30] T. H. Murray. Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different from Other Medical Information? in: M. A. Rothstein (ed.), Genetic Secrets. Protecting Privacy and Confidentiality in the Genetic Era, New Haven/London: Yale University Press; 1997: 60-73; D. B. Paul. What Is a Genetic Test, and Why Does It Matter? in: Endeavour. 1999; 23: 159-161, here: p. 160; European Commission, op. cit. note 26, p. 44.
[31] See on the following J. Beckwith, J. S. Alper op. cit. note 29, p. 143-144.
[32] G. Feuerstein, R. Kollek. Risikofaktor Prädiktion. Unsicherheitsdimensionen diagnostischer Humanexperimente am Beispiel prädiktiver Brustkrebstests, in: L. Honnefelder/C. Streffer (eds.), Jahrbuch für Wissenschaft und Ethik, 5. Berlin: De Gruyter; 2000: 91-115; T. Lemke. Veranlagung und Verantwortung. Genetische Diagnostik zwischen Selbstbestimmung und Schicksal, Bielefeld: transcript Verlag; 2004: 70-76.
[33] A. Kerr. (Re)Constructing Genetic Disease: The Clinical Continuum between Cystic Fibrosis and Male Infertility, in: Social Studies of Science. 2000; 30: 847-894; H. van den Boer-van den Berg, A. A. Maat-Kievit. The whole truth and nothing but the truth, but what is the truth? in: Journal of Medical Genetics. 2001; 38: 39-42.
[34] R. Hubbard, R. C. Lewontin. Pitfalls of genetic testing, in: The New England Journal of Medicine. 1996; 334, S. 1192-1194; U. Wolf. Identical mutations and phenotypic variation, in: Human Genetics. 1997; 100: 305-321.
[35] Murray op. cit. note 30, p. 64; R. L. Zimmern. Genetic Testing: a Conceptual Exploration, in: Journal of Medical Ethics. 1999; 25: 151-156; European Commission op. cit. note 26, p. 44.
[36] Murray op. cit. note 30. p. 65.
[37] While today a hard-line genetic determinism is less accepted in genome research, we can still observe "the narrative of enlightened geneticization", that presents current genetic thinking as non-reductionist, grants a role for non-genetic factors while at the same time prioritizing genetic explanations to behavior or disease (A. Hedgecoe. Schizophrenia and the Narrative of Enlightened Geneticization, in: Social Studies of Science. 2001; 31: 875-911; T. Lemke. Mutationen des Gendiskurses: Der genetische Determinismus nach dem Humangenomprojekt, in: Leviathan. Zeitschrift für Sozialwissenschaft. 2002; 30: 400-425).
[38] E. F. Keller. The century of the gene, Cambridge, MA/London: Harvard UP; 2000; E. F. Keller. Making Sense of Life. Explaining Biological Development with Models, Metaphors, and Machines, Cambridge, MA/London: Harvard UP; 2002.
[39] Developmental systems theory shows that a rigid juxtaposition of culture and nature does not suffice to explain biological processes. It assumes that biological characteristics refer by no means to a culturally-independent and trans-historical essential core; they are the result, not the cause of developmental processes within a complex system, in which social and psychological factors also play a key role. This research program thus severs any ties with biological or social variants of determinism (cf. S. Oyama. The Ontology of Information, Duke University Press; 2000; see also A. Fausto-Sterling. The problem with sex/gender and nature/nurture, in: S. J. Williams, L. Birke, G. A. Bendelow (eds.), Debating Biology. Sociological reflections on health, medicine and society, New York/London: Routledge; 2003: 123-132).
[40] I shall return to this point below.

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[41] Murray op. cit. note 30, p. 65-66; J. S. Geetter. Coding for Change: the Power of the Human Genome to Transform the American Health Insurance System, in: American Journal of Law & Medicine. 2002; 28: 1-76.
On the relationship between health and responsibility, see S. J. Reiser. Responsibility for Personal Health – a Historical Perspective, in: Journal of Medicine and Philosophy. 1985; 10: 7-17; A. M. Brandt, P. Rozin (eds.). Morality and Health, New York & London: Routledge; 1997.
[42] See L. E. Kay. Who wrote the book of life? A history of the genetic code, Stanford: Stanford University Press; 2000; L. Moss. What genes can't do, Cambridge/MA und London: MIT Press; 2003.
[43] E. J. Yoxen. Constructing Genetic Diseases, in: T. Duster, K. Garrett (eds.), Cultural Perspectives on Biological Knowledge, Norwood/NJ: Ablex Publishing Corporation; 1984: 41-62; T. Lemke. Molekulare Medizin? Anmerkungen zur Ausweitung und Redefinition des Konzepts der genetischen Krankheit, in: Prokla. Zeitschrift für kritische Sozialwissenschaft. 2003; 33: 471-492.
Murray talks of a "two-bucket theory of disease" that in the final instance rests on the erroneous assumption that we can medically distinguish clearly between genetic and non-genetic illnesses or factors for illness: "According to this model, there are two buckets – one labeled 'genetic', the other labeled 'nongenetic' – and we should be able to toss every disease and risk factor into one of the two. So, Huntington disease goes into the 'genetic' bucket and getting run over by a truck goes into the 'nongenetic' one. But many diseases and risks don't fit neatly into either bucket. Take breast cancer. Some cases of breast cancer have strong genetic roots, but others have no clear genetic connection. For that matter, not every woman with a mutated BRCA1 gene will develop breast cancer. And some apparent risk factors have little or no link to genetics. Similar complexity exists for heart disease: cholesterol is a risk factor, and one's cholesterol level can be modified by diet, exercise, and other factors; but our genes have as much or more to do with the level of cholesterol circulating in our blood as our environment or behavior. Into which bucket, then, should we toss breast cancer? Heart disease? Cholesterol level?" (Murray, op. cit. note 30, p. 67-68).
[44] Zimmern op. cit. note 35; J. Schmidtke, K. Sperling. Genetische Tests auf dem Teststand, in: Zeitschrift für Biopolitik. 2003; 2: 39-47.
[45] Alper/Beckwith, op. cit. note 29, p. 145. J. Beckwith, J. S. Alper. Reconsidering Genetic Antidiscrimination Legislation, in: Journal of Law, Medicine and Ethics. 1998; 26: 205-210, here: p. 207.
Zimmern has pointed out that the concept of genetic information possesses two mutually contradictory meanings: "First it may be regarded as information about the genetic constitution of individuals, their genes or chromosomes, and their inheritance. Second, and by contrast, genetic information may be taken to refer to any information from which we may infer knowledge about the genetic constitution of individuals" (op. cit. note 35, p. 152).
[46] Paul, op. cit. note 30. On the history of PKU see D. B. Paul. PKU Screening: Competing Agendas, Converging Stories. in: D. B. Paul, The Politics of Heredity. Essays on Eugenics, Biomedicine, and the Nature-Nurture Debate, Albany: State University of New York; 1998: 173-186.
[47] Billings et al., op. cit. note 3.
[48] See, for example, M. Rothblatt. Unzipped Genes. Taking charge of baby-making in the new millenium, Philadelphia: Temple University Press; 1997: 157: "If we have a genomic predisposition to a particular condition that interferes with our job ability, then it is wrong to deny that job until the condition manifests itself."
[49] G. Wolbring. Folgen der Anwendung genetischer Diagnostik für behinderte Menschen, Berlin: Gutachten erstellt im Auftrag der Enquete-Kommission des Deutschen Bundestages "Recht und Ethik der modernen Medizin"; 2001: 87.
[50] F. Mechan-Schmidt. Teacher protests at gene bias, in: Times Educational Supplement, 14th november 2003; J. Burgermeister. Teacher was refused job because relatives have Huntington's disease, in: British Medical Journal. 2003; 327: 827-a; G. Traufetter. Geisel der eigenen Gene, in: Der Spiegel. 2003, no. 42: 216-218.
[51] Huntington's Disease is a neurodegenerative illness that usually breaks out in the fourth or fifth decade of the person's life. It is triggered by an increased repetition of a certain triplet (the sequences of three nuclear bases cytosine, adenine, guanine: CAG) in the Huntington gene, which is located on the short arm of Chromosome 4 (cf. H. W. Lange. Morbus Huntington – Klinik, Diagnose und Therapie, in: Psycho. 2002; 28: 479-486).
[52] Frankfurter Rundschau. Erblich belastete Lehrerin wird Beamtin, in: Frankfurter Rundschau, 3rd August 2004; Ruling of the Darmstadt Administrative Court of June 24, 2004, AZ 1 E 470/04 (3).

[53] Frankfurter Rundschau. Zu dick für Beamtenjob, in: Frankfurter Rundschau, 3rd june 2004.

[54] Alper/Beckwith, op. cit. note 29, p. 147; Zimmern, op. cit. note 35, p. 153.

[55] See Geetter, op. cit. note 41.

[56] A legal prohibition on genetic discrimination would probably lead to further commercialization of the genetic diagnostics sector or enable this to happen in the first place. For example, William Rice, President and Medical Director of myDNA, the US provider of genetic diagnostics, expressly champions a federal anti-discrimination law, as he believes most citizens will in the absence of protection of their genetic privacy avoid genetic screening in order to prevent discrimination in the place of work and in insurance contracts (Yahoo Financial News. My DNA Media Endorses Passage of the Genetic Non-Discrimination Act of 2003, in: Yahoo Financial News, 21st june 2004.). In Germany, the Diagnostics Industry Association issued a similar statement in a communiqué that advocates supporting the planned genetic diagnostics law (see K.-P. Görlitzer. Gentest-Gesetz nach Wünschen von Kassen, Forschern und Firmen, in: Bioskop. 2004; 7 (28): 8-9).

[57] Cf. E. R. Shell. The Hungry Gene. The Science of Fat and the Future of Thin, New York: Atlantic Monthly Press; 2002; J. Hebebrand. Mehr Übergewicht – mehr Krankheiten? Genetische Ursachen der Übergewichtigkeit werden erforscht, in: GenomXPress. 2004; no. 4: 19-21.

[58] See on this also Wolf, op. cit. note 24.

[59] This argument is also typical of many scholarly articles that critically focus on practices of genetic discrimination. See, for example, L. Gostin. Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers, in: American Journal of Law & Medicine. 1991; 17 (no. 1+2): 109-144, p. 110-1: “Prejudice, alienation and exclusion often accompany genetically related diseases even though, by definition, the condition is neither subject to the person’s control, nor the result of willful behavior.” Likewise, Rothblatt, op. cit. note 48: 157: „Genomic discrimination is wrong because it categorizes us on the basis of biology over which we have no control and which is irrelevant for any social, economic, or legal purpose.“

[60] Bundesministerium für Gesundheit und Soziales. Begründung zum Diskussionsentwurf “Gesetz über genetische Untersuchungen bei Menschen”, Berlin; 200: 16. For a similar point see the statement of the German Minister of Justice who holds that genes are “the most valuable good that a human being possesses.” (cited by C. Schwägerl. Gentests. Prinzip Selbstbestimmung, in: Frankfurter Allgemeine Zeitung, 14th Januar 2005: 4).

[61] Diver/Cohen, op. cit. note 14, p. 1451-1452.

[62] See also Beckwith/Alper, op cit. note 45, p. 208: “This rationale seems to carry with it the invidious implication that we are responsible for our nongenetic diseases.”

[63] H. Kühn. “Selbstverantwortung” in der Gesundheitspolitik, in: Jahrbuch für kritische Medizin. 1998; no. 30: 7-20; M. Bause. Guter Rat ist teuer – humangenetische Beratung unter den Bedingungen der Marktindividualisierung, in: J. Schmidtke (ed.), Guter Rat ist teuer. Was kostet die Humangenetik, was nutzt sie? Munich & Jena: Urban & Fischer; 2000: 96-106.

[64] Ingrid Schneider points to a similar problem in the context of the debate on patenting human genes: “However, because critics and the persons affected assert that the patenting robs them of their innermost being or ‘identity’, they reproduce and confirm a genetic determinism and essentialism that erroneously reduces human existence to DNA structures” (I. Schneider. Patente Argumente? in: Bioskop. 1999; no. 8: 14-15, p. 15).

[65] See, among others, Beckwith/Alper, op. cit. note 45, p. 208.

[66] This argument is explained in detail by D. Hellman, op. cit. note 11, p. 79: “This article will develop the argument that because the social meaning of treating people differently on the basis of their genetic make-up is different from the social meaning of discrimination on the basis of health or illness, special legislation is warranted to prohibit genetic discrimination”; see also Murray, op. cit. note 30, p. 71.

[67] See the critical analysis in D. Nelkin, S. M. Lindee. The DNA Mystique. The Gene As a Cultural Icon, New York: W. H. Freemann & Co; 1995; J. van Dijck. Imagenation. Popular Images of Genetics, New York: New York University Press; 1998.

[68] J. Schmidtke. Vererbung und Ererbtes – Ein humangenetischer Ratgeber, Reinbek: Rowohlt; 1997: 13.

[69] T. Burnham, J. Phelan. Mean Genes. From Sex to Money to Food: Taming our Primal Instincts, Cambridge/MA: Perseus Publishing; 2000; P. Little. Genetic Destinies, Oxford: Oxford University Press; 2002.

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[70] A. M. Kavanagh, D. H. Broom. Embodied Risk: My Body, Myself? in: Social Science & Medicine. 1998; 46: 437-444.
[71] V. Senior, T. M. Marteau, T. J. Peters. Will genetic testing for predisposition for disease result in fatalism? A qualitative study of parents responses to neonatal screening for familial hypercholesterolaemia, in: Social Science & Medicine. 1999; 48: 1857-60.
[72] N. Holtzman, M. A. Rothstein. Invited Editorial – Eugenics and Genetic Discrimination, in: American Journal of Human Genetics. 1992; 50: 457-459; D. J. Kevles. In the name of eugenics: genetics and the uses of human heredity. Cambridge, MA/London: Harvard University Press; 1995; H. Markel. The Stigma of Disease – Implications of Genetic Screening, in: American Journal of Medicine. 1992; 93: 209-215; P. Weingart, J. Kroll, K. Bayertz. Rasse, Blut und Gene. Geschichte der Eugenik und Rassenhygiene in Deutschland, Frankfurt am Main: Suhrkamp; 1992.
[73] Alper/Beckwith op. cit. note 29, p. 148; Beckwith/Alper, op. cit. note 45, p. 209; Geetter, op. cit. note 41.
[74] See Wolf op. cit. note 24.

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New mothers' awareness of newborn screening, and their attitudes to the retention and use of screening samples for research purposes

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Abstract

Aim: To explore new mothers' knowledge of newborn screening, and their attitudes towards issues surrounding sample retention and the potential for blood screening samples to be used for research.

Methods: A self-administered mail survey was sent to women who gave birth in Perth, Western Australia during January 2005. A total of 600 women completed the survey.

Results: It was found that women were aware of newborn screening, however desired further information in order to acquire a more comprehensive knowledge of the test. Further, women reported discomfort with the long-term storage of cards, but they were supportive of using blood samples for medical research, contingent upon the samples being de-identified and parental consent provided.

Conclusions: New mothers need to be provided with comprehensive information about the newborn screening test at a time which is conducive for the assimilation of this information. In addition, whilst supporting health related research using newborn screening samples, new mothers are keen for ethical issues to be sufficiently addressed prior to samples being systematically stored for extended periods of time.

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Introduction

For several decades the genetic blood screening of newborns to detect inborn errors of metabolism has been recognised as a valuable component of neonatal care in many developed countries around the world.[1 2] The early detection of these disorders has proven an effective means by which interventions can be implemented to significantly reduce morbidity, mortality and associated disabilities.[3 4 5] Publicly funded newborn screening programs have been operating in Australia since 1964, and although participation is entirely voluntary, there is high public participation in most programs1, enabling them to be cost effective.[6]

At present Australian newborn screening programs are working towards developing a nationally consistent approach to the retention of newborn screening cards, and secondary uses of the blood samples derived from these cards. At the time of this study in 2005, retention periods still vary significantly across States, ranging from two years to indefinitely. Such considerable differences between programs is reflective of the situation internationally where there is no agreement on the appropriate retention period of newborn blood samples.[2,7] Closely associated with sample retention periods is the issue of what, if any, secondary uses are appropriate for the blood samples collected for the sole purpose of newborn screening.

While sample retention for forensic and quality assurance purposes, and for the development or modification of screening tests is justifiable, it is the potential for these samples to be used to conduct population based health screening studies and epidemiological research that prompts the need to articulate agreed standards in this area. It is very difficult to obtain population datasets of this kind that derive from the whole population with no selection.[7] Consequently, the research opportunities are plentiful. In the United Kingdom, the value of conducting research based on blood samples gathered through newborn screening is acknowledged as having contributed towards answering important public health questions and leading to advancements in newborn and antenatal screening technologies.[8]

At the same time, significant ethical issues underlie the ability to access blood samples derived from newborn screening, including the appropriateness of using samples obtained through dissent rather than informed consent for secondary purposes,[2] and the ability to garner public support for such research. Biological specimen databanks are often met with great reservation by the public because of their potential for misuse and a lack of visible bodies to provide regulation.[7] Further, these ethical issues may vary according to the levels of access researchers would be afforded, in particular whether they would have access to identified or de-identified samples.

Ascertaining the views of the community, and parents in particular, about the retention and use of newborn blood samples forms a critical component of the development of any policy in this area. Such studies are a growing area of investigation within the psychology of newborn screening,[9] although not yet explored extensively. To date these studies have targeted the parent's levels of knowledge about the programs and any psychosocial issues related to the provision of positive test results for particular disorders.[10 11 12 13] It is reasonable to anticipate that the attitudes of parents whose

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child has been diagnosed through newborn screening may be different from those parents whose child was found not to have an inborn error of metabolism, or from the community in general.

Although several factors influence parental views about newborn screening, parents have been found to be generally supportive of newborn screening programs, even in the case where false-positive test results have been given.[4] Positive attitudes are not necessarily contingent upon adequate knowledge of screening, as there is substantial evidence suggesting that parents often have limited knowledge of which disorders are tested for, the effects of the disorders and the treatments available.[9] Many new mothers are not even aware of the test being performed on their child.[14] Further, a recent study has found that parents in the United States are not well informed about the storage of cards, or their potential uses, and only five percent of educational materials aimed at informing parents about newborn screening actually address these issues.[15] Although data regarding attitudes towards storage and use of newborn screening samples is limited, Gustafsson Stolt et al (2002) report their respondents expressing concern about the storage of material and the right to be informed of any screening or project results.[16]

In relation to using newborn blood samples for research purposes, data from Sweden suggests that mothers have generally positive attitudes to research. Those who choose to allow their child's blood sample to be used for research cite the potential to contribute to research as the primary motivation for doing so.[16] Those who do not allow participation often cite concerns about making decisions on behalf of their child regarding genetic material as a primary consideration.[16]

The paucity of information about parental attitudes in relation to issues in newborn screening,[13] has been identified as an issue requiring exploration. For policy development in particular, greater dialogue between government and community is necessary so that community concerns and any associated ethical issues may be adequately addressed.[7,16] This study aims to explore new mothers' knowledge and attitudes towards newborn genetic blood screening. Specifically, it aims to ascertain new mothers' awareness of newborn screening, and their attitudes about issues relating to the retention and use of blood samples to include research purposes in Western Australia.

Methods

Participants

Women who gave birth during the month of January 2005 were invited to participate. Those women who gave birth to a stillborn child or those whose child died neonatally were excluded.

Measures

Information was collected via a self-administered mail survey, which women received four month's after the birth of their child. Items included in the survey were constructed following a review of the literature. Further, the investigators met with a group of new mothers to ask them about their experience with newborn blood screening to inform survey construction. A pilot study was also conducted to verify ease of understanding.

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The survey investigated four key areas, namely awareness of newborn screening, attitudes about appropriate retention periods for samples, attitudes towards the use of samples for secondary purposes specifically research, and demographic information. Participants were also given the opportunity to provide comments in order to enable aspects of these issues, which could not be explored through structured questions to be investigated.

Procedure

Women meeting the inclusion criteria were selected from the Midwives Notification Database located at the Department of Health (Western Australia). These women were sent a copy of the survey, along with an information sheet detailing the method of participant selection, the purpose of the study, background information on newborn screening, and a reply paid envelope. Women were informed that completion and return of the survey was deemed consent to participate.

The University of Western Australia Human Research Ethics Committee approved the study and the Confidentiality in Health Information Committee at the Department of Health, Western Australia granted access to the Midwives Notification Database.

Data Analysis

Descriptive statistics, Spearman's Rank Order Correlation multiple response analysis procedures were used to analyse the data.

Results

The survey was sent to 1846 new mothers, of which 600 returned a completed survey, equating to a response rate of 33%. Although this response rate is typical for a self-completed mail survey,[17] and for new mothers in particular,[18] an independent group of new mothers was sampled through seven new mother's groups in the metropolitan area to check for bias in the study cohort. All new mothers present completed a short version of the mail survey (N = 52). Demographic data were not collected on these new mothers. Responses from this sample were consistent with those obtained in the main survey.

Ages ranged from 18 to 47 years with the mean age of the sample being 32 years. The number of children the women had ranged from one child to seven, with the average being two children. First time mothers comprised 44% of the sample. The majority of women lived within the metropolitan area (78%). Over a third of women had a university education (36%), and 27% reported that they had post-secondary school qualifications.

To determine the representativeness of the sample, data from the Midwives Notification Database on all women who gave birth during January 2005 was obtained. Table 1 illustrates the differences between the population group and the sample collected.

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Table 1. *Comparison of population and sample demographics*

	Population (N = 1945)	Sample (N = 600)
Mean age	29.4 years	31.9 years
Mean number of children	1	2
Geographical area		
Metropolitan	74%	78%
Rural	26%	22%

Awareness of Newborn Genetic Blood Screening

The majority of women (93%) stated that they had heard of newborn genetic blood screening prior to receiving the survey. They reported receiving this information from a variety of sources (Table 2).

Table 2. *Respondent's reported sources of information about newborn screening*

Sources of information	Most popular sources reported (%)
Midwife	34%
Previous pregnancy	27%
Newborn screening pamphlet	14%
General practitioner or	12%
obstetrician	12%
Family or friends	1%
Internet	

Over half of all respondents were satisfied with the information provided (51%), with the remainder reporting higher (19%) or lower levels of satisfaction (18%). When provided with the opportunity to comment further, the women stated that the time at which the information was provided was a significant factor in determining their ability to adequately consider the information:

> *I feel the test isn't discussed enough at the time it is done. A midwife takes your baby, does the test, brings baby back and leaves a pamphlet that on most occasions gets put aside with all that's going on.*

> *(The information) could have been lost in avalanche of other information and emotion.*

Women also reported that they would have liked to receive more comprehensive information:

> *Only very general information was given to me when the tests were done. I would have liked more detailed information (from midwife or information booklets).*

> *I don't believe the information I received from my GP, Obstetrician, or hospital was adequate.*

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In order to gauge the extent to which women valued newborn genetic blood screening, they were asked to rate their agreement with the following statement: '*I believe newborn blood screening is valuable for enabling the early detection of genetic disorders in children*'. The majority either strongly agreed (61%) or agreed (34%). Only 5% stated that they either disagreed or held no opinion. Women who had received information about newborn screening are more likely to value the screening test (.099, p = .017) and women's belief in the value of newborn screening positively correlated with the degree to which they are satisfied with the information they were provided (.370, p = .000).

Newborn Blood Sample Retention Periods

Women were asked to nominate an appropriate time-period for retaining newborn blood samples before they are destroyed (Table 3).

Table 3. *Support for proposed newborn screening blood sample retention periods*

Period of Retention	Percentage of Respondents
2 years or less	29.3%
3 – 10 years	29.5%
11 – 20 years	8.6%
21 years or more	16.8%
Indefinitely	0.6%
Unsure	6.3%
As long as they are required	8.9%

When asked to nominate reasons for their choice, women who believed in maintaining the retention period of two years spoke of not possessing adequate knowledge of the issues involved to justify deviating from current practice:

> *I do not know a lot about the screening test nor have heard of reasons why they may be kept longer. Perhaps if I had more information my answer may be different.*

> *I don't know enough about the information gathered from the screening to believe the timeframe should be different from the present period of two years.*

Other reasons for maintaining the current practice two-year retention period included statements that the time was adequate for the primary purpose of the test to be achieved and that this short period of time is a safeguard against any unnecessary research:

> *My understanding is the disorders checked for occur in infancy…and two years should be adequate to gather data for any research needs.*

> *(Two years is appropriate) so information is gathered promptly and cards don't just sit there waiting for someone to come up with something new to research.*

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Some women also distinguished between the results of the tests, believing that a longer period of retention could be justifiable for those samples, which test positive to enable research.

Women who nominated between 3-10 years as being an appropriate retention period were keen to promote research on the samples, believing that this time enabled more research to take place. They also reported the belief that this period allowed for advancements in technology to enable the cards to be re-checked if necessary:

> *It could provide a reference point if the child develops any problems beyond two years, plus it may be useful for future research which should be automatic unless parent specifies otherwise.*

The longer retention periods of cards was also nominated because it is in keeping with standard record retention periods of between 5 to 10 years for important documentation, such as those required for taxation purposes.

There was also support for a retention period of 21 years or longer (17%), the advantage of which was the facilitation of research opportunities.

Attitudes towards the Use of Newborn Blood Samples for Research Purposes
Most of the cohort (85%) believed that de-identified newborn screening samples should be made available for research, the remainder either disagreed (4%) or were unsure (11%).

In order to gain an appreciation for the strength of women's belief in the use of cards for research, they were asked the extent to which they agreed with the statement "*I would agree to my baby's card being used for research*". It was found that 85% of respondents either strongly agreed or agreed with the statement. A further 9% held no opinion, and 5% disagreed. In addition, 79% either strongly agreed or agreed with the statement "*I would like to have the opportunity to contribute positively to research through newborn screening cards*". This sense of support for research was also reflected in the comments:

> *Whatever information can be collected and research done on cards can only be of a benefit to medical community to better understand genetic diseases or other diseases that are influenced by lifestyle / environment.*

The women viewed de-identification of blood samples as a priority. It was found that when questioned about the importance of de-identifying blood samples for any purpose other than newborn screening, 90% of the sample agreed that this should occur. The importance of de-identification was also demonstrated through the comments provided:

> *I am happy for my child's blood screening test to be used for research…as long as my child's identity and personal information is removed.*

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> *I feel strongly about ensuring that genetic details and information is kept private. Guaranteed privacy of genetic information would be essential to my support of any research.*

Closely associated with this is their belief that parental consent should be sought prior to the samples being made available for research purposes:

> *I feel strongly that consent should have to be given before samples are used for any purpose other than the specified genetic tests.*

> *Additionally, women were keen to receive information about the types of research that may be conducted so that they could make an informed decision as to whether the research is compatible with their ethical values:*

If this information was used for research I would like to know what type of research to decide if I agreed with it ethically and morally.

I would be fine with use of sample for research if it was specifically outlined what the use would be and how the sample would be stored / destroyed / managed.

In order to ascertain if women's attitudes would be dependent upon the type of research that the samples could be used for, four research areas were listed and women were asked to nominate the extent to which they felt using the samples would be appropriate. These areas were (i) to understand the prevalence of disease in the community, (ii) to improve diagnostic tests for childhood diseases, (iii) to understand how lifestyle factors influence genetic diseases, and (iv) to improve diagnostic testing of diseases. All four areas were overwhelmingly supported. The most support was given to improving diagnostic tests for childhood diseases (97%), followed by improving diagnostic testing (96%), understanding prevalence of disease (93%), and lifestyle and genetic interactions (92%).

When asked to nominate any types of research that would be unacceptable uses of newborn screening cards, the fields of uses most commonly cited were cloning, research that leads to abortion, 'designer babies', and the use of the information for paternity issues or criminal investigations.

Further, women noted the need for necessary safeguards against inappropriate use to be in place prior any changes to current practice:

> *I would be happy for my child's card to be used for research but have concerns regarding the safety of personal and genetic information, and the relevance of the research. I wouldn't want the cards kept for the purposes of doing research that does not directly benefit the community.*

> *Ethics is an important issue here and the availability of data to insurance companies, other family members, police etc without consent or by law is a problem not yet dealt with.*

Other Relationships

No relationships between demographic factors and outcomes on the key variables were found.

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Discussion

This study investigated knowledge and attitudes of new mothers who had received no further information about newborn screening from the researchers, other than that presented in the newborn screening pamphlet, prior to completing the survey. In this way, the results generated provide a sound indication of the views of new mothers in Western Australia about issues relating to newborn screening and the potential use of screening samples for secondary purposes.

The results suggest that women are aware of newborn screening, but do not feel that they are well informed. This echoes previous research indicating that new mothers possess limited awareness of the newborn screening test being performed,[14] and its purpose, including the diseases being tested.[9,19] Most women received screening information within the 72-hour period after the birth of their child. During this time, they receive a plethora of information relating to other aspects of neonatal care. It is also common for midwives to perform the blood test away from the mother to circumvent any unnecessary anxiety,[14] so it is not surprising that women may have difficulty assimilating screening information for a test for which they must 'opt-out'.

Previous studies have shown that the time at which screening information is provided was a critical determinant of women's ability to adequately absorb the information. Therefore it may be more beneficial for health professionals to endeavour to provide this information during the prenatal period.[2,15] In addition to facilitating the comprehension of the material, presenting information at this time would also enable more detail to be passed onto parents, and provide an opportunity for questions to be asked. Women were keen to receive additional newborn screening information through government pamphlets. Only a small proportion of women reported reading this pamphlet, and therefore it may be useful to ascertain if other forms of communicating this information may be more relevant for this group.

Ensuring that women have adequate time to consider screening information becomes even more salient when considering the possibility of research on newborn blood samples. There was considerable support for the use of blood samples for research, and this support was motivated by the desire to make a positive contribution to research, mirroring results found for a similar cohort overseas.[16] The women were also particularly supportive of research targeted at child health issues, indicating a belief that research on stored samples should be closely aligned with the primary purpose of the sample collection. However, support for research was contingent upon safeguards against breaches to privacy and policy makers need to be mindful of the public's wariness of biobanks and ensure that the appropriate safeguards are in place prior to the systematic storage of newborn blood samples for extended periods.

In relation to retention periods, the strong support of maintaining the current WA storage period of two years is to be expected. Without a thorough knowledge of the issues, determining a justification for nominating a longer time-period is difficult. Nevertheless, the women equally supported storage of between three to ten years to allow adequate time for research. The qualitative data also suggests that women are generally uncomfortable with the long-term retention of cards for research purposes. These results suggest that given appropriate justification, samples may be stored for

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longer than the current two-year period, although storage beyond a decade is unsupported.

There were specific aspects arising from this research which merit further investigation. In particular, this study did not collect information on whether any of the women participating had received either a positive or a false positive test result. Although the expected numbers for such results would be low, it would nevertheless be valuable to explore any differentiation between these groups from the core group across all variables. Secondly, the investigation of women's awareness of screening could be explored in greater detail to gain an indication of women's actual knowledge levels, for example, their knowledge of what conditions are screened for in testing.

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Health as a genetic planning project: Enthusiasm and second thoughts among biomedical researchers and their research subjects

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Abstract

This paper presents an interview study among scientists working with Decode genetics in Iceland and lay individuals having recently donated blood to Decode. While genuinely enthusiastic that genetic technologies hold great potential to avert disease, the informants shared concerns that extensive predictive genetic testing, preventive treatment and tailoring of lifestyle to avoid potential disease may cause loss of freedom – people can "worry themselves sick". Undiscriminating use of genetic technologies in privileged populations was seen as a potential source of injustice and reduced tolerance of diversity. Both lay informants and scientists revealed ambiguity and inconsistency in their personal evaluation of genetic knowledge, indicating that 'rational choice' models do not predict how people relate to information about risk, expert knowledge notwithstanding. Drawing on work by Wynne and van Hooft, we submit that our informants' ambivalence and second thoughts are implicit contradictions of prescriptive messages accompanying human genetics – i.e. more or less covert and non-intentional claims about the rational obligation to minimise the likelihood that one falls ill and a strictly biological conception of health. Genetic technologies designed to prevent or combat organic disease can interfere negatively with non-biological levels of health. It is a challenge of reflexive modernity to untangle the interaction of human genetics with culturally mediated categories of relatedness, purpose and meaning in everyday life, and mobilise cultural and governance resources which can ensure that genetic technologies support human subjectivity and health in their full range.

Introduction

Public debate on human genetics can be both polarized and polarizing. On one hand, a significant subset of the public in many countries is wary of many uses of human genetics and certain other biotechnologies. On the other, dominant science policy discourse – apparently supported by the majority of the public – expresses a general approval of these technologies as a major source of improved health, quality of life and economic growth, with a few exceptions such as human reproductive cloning.[1] Mass media appear to reinforce polarization by their very repertoire in the framing of human genetics. There is the frame of technological optimism, emphasising scientific bravery and reproducing scientific promises of relief from disease and suffering.[2] Alternatively, if the technology in question is somehow controversial (such as reproductive cloning), a frame of fear and horror is easily applied, evoking the Frankenstein myth, as it were.[3] A similar polarization tends to be conjured by the expert and media portrayal of public sentiments, as in the case of GM food, held by British media and expert reports to divide the public into mutually exclusive fractions.[4]

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Nevertheless, contemporary societies arguably have made the transition into reflexive modernity, in which citizens recognize and debate the joint production and redistribution of benefits and risks as an inherent aspect of scientific and technological advance. Hence, people's sentiments towards genetic technologies are likely to be more nuanced, reflexive and ambivalent than what is conveyed by policy debate, mass media or even by surveys.[5] The marginalization or privatization of reflexivity may in part be due to the dominance of technological optimism in the policy discourse about healthcare and genetic research. Despite evidence about the shortcomings of the so-called 'deficit model', second thoughts and reservation regarding genetic technologies are frequently de-legitimised in public debate by blaming them on ignorance. But marginalization may also be an artefact of reporting, as mainstream media rendering of quantitative surveys does not easily capture complex reasoning, nuanced sentiments or ambivalence.

This paper presents an exploration of reflexivity through a qualitative study among individuals knowledgeable and *prima facie* supportive of human genetics in Iceland. We believe this task to be important. Discrepancies between lay and expert perception of human genetic technology, and more subtly, between real and presumed perceptions, may easily translate into an erosion of public legitimacy similar to that which has shaken the governance of related technologies in recent years.[6] Further, unacknowledged reflexivity will not feed into processes of governance, increasing the danger that technology will cause more harm than benefit. This raises the need to understand the public understanding of genetics as well as the democratic challenges of developing inclusive governance taking that understanding into account.

In Iceland, the private enterprise Decode genetics has collected data from more than 50% of the adult population for their research into the genetic components of many common diseases, including myocardial infarction, schizophrenia and asthma. Following a fierce debate locally and internationally in 1997-2000 about Decode's plans for a nation-wide database with comprehensive healthcare information, the company recently appears to have achieved a favourable position in the Icelandic social and cultural imagery.[7] According to a Eurobarometer conducted in 2005, the Icelandic public is more supportive of biobank research and biotechnology than people in any other European country,[8] and according to the company itself more than 90% of the public respond favourably when asked to contribute to Decode's research projects.[9] Our hypothesis was that eliciting reflections in this setting, among individuals knowledgeable and supportive of human genetic research, would allow a philosophical-anthropological analysis of different and potentially conflicting meanings attached to genetic technologies, in particular with respect to personal health risk management. Accordingly, we performed focus group interviews about the impact of human genetics on everyday life with scientists working with Decode genetics, as well as with individuals who recently had contributed to Decode's research, i.e. donors of blood samples and personal information.

This study forms part of a larger investigation into the reactions to Decode genetics and the governance of human genetics in Iceland.[10] It is our opinion that the challenge of reflexive modernity is to pass into *pre hoc* anticipation of higher-level effects of genetics to improve the governance of novel technologies. By 'higher level effects' we mean the socio-cultural repercussions of genetic research and the associated

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technologies. Before presenting and discussing the results of our focus group interviews, we will outline a general framework for the higher-level effects of science and technology as found in the work of Brian Wynne, followed by a brief account of Stan van Hooft's theory of health, which we take to provide a useful conceptualisation of some of the most central dimensions of culture interrelated with the current rise of human genetic research.

Theoretical framework

Wynne writes that public reactions to science and technology concern not only the concrete facts, proposals and applications, but also the 'prescriptive messages' of technological projects.[11] These are value-laden messages about human agency, the nature of human existence, the role of science in the broad context of everyday life, etc., that accompany science and technology as their presuppositions or implications. The transmission of such messages is a hermeneutic process, resulting not only from more or less deliberate intentions originating in expert institutions, but also through a dialectic of lay interpretations, reflecting a variety of knowledge, values, fears and hopes. According to Wynne, public interpretation of the prescriptive models of human agency and risk management is germane to the public critique of science, as in the case of, say, hostility towards GM food and crops and technological solutions to environmental pollution.[12]

Following Wynne's line of reasoning, the subtleties of public perceptions of human genetic technologies are directly relevant to the implications of these technologies on health and health care. Already from Thomas' and Thomas' classic theorem, "If men define situations as real, they are real in their consequences",[13] it follows that public perception will feed back into real action in society. In particular, we expect the practical lifeworld consequences of these technologies to be broader than the face-value clinical utility, and to depend upon the ways in which they are understood and integrated into thoughts, expectations and actions. This integration is crucially linked with what Wynne calls prescriptive messages, and will depend upon a large number of factors including scientists', stakeholders' and media's appearance in terms of trustworthiness, competency and responsibility. However, integration also has to be seen as an essentially open-ended and unpredictable process in which the public and society perform a creative translatory work upon the technologies, their prescriptive messages, and their own world-views and value systems.

A central lifeworld dimension involved in the rise of human genetics is health itself. By promising and providing new knowledge about disease mechanisms, risk, and the potential consequences of a person's attitudes towards genetic tests, lifestyle and reproduction, genetics unleashes an unlimited number of intended and unintended 'messages' which interact with people's thoughts, actions, relationships and identities. Conversely, contemporary beliefs about human agency, disease and the power of medical interventions form the cultural environment which enables and gives shape to the rapid development of genetics. To deal with certain features of this interaction, we shall apply the broad concept of health developed by van Hooft, in which health is seen as inseparable from human subjectivity and as such inherently value-laden and conditioned by the particularity of individual persons, embedded in the language and culture of late modernity.[14] The crucial feature of van Hooft's account in our context

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is that it expands the biomedical model of health by identifying four levels of subjectivity, referring to different aspects of human existence, from molecule to the meaning of life, where suffering and loss of health may arise. The first level is the biological and concerns the functioning of organs, tissues and molecules. The second, relational, level denotes the ways in which we relate to reality through perception and interpretation, depending on largely unconscious processes of meaning-construction. The third, pragmatic, level concerns people's ability to fulfill social roles and plan their lives. The fourth level, integration, denotes our need for self-understanding, purpose and belonging, our dependence on community and narratives that help us commit ourselves and see life as worthwhile. Although the three non-biological levels of health can be distinguished from each other for analytical purposes, in reality they tend to be integrated with each other and with the biological dimension. For our present purpose, distinguishing the non-biological dimensions of health from biological health will be crucial. It is easy to see that activities focusing solely on one level may result in unanticipated effects on a different level. Indeed, the history of medicine bears evidence of numerous instances where medical interventions effectively targeting the biological level also had unpredicted health effects on the three other levels. Certain quarantine and eugenic practices are but two examples of practices that were abolished not primarily because they were ineffective or harmful to health on the biological level, but because they were found to have detrimental effects on the three other levels. Also, a significant reason for the current rise of alternative therapies in societies with advanced biomedical healthcare services seems to be the tendency of biomedical practices to focus on the biological, while not adequately addressing higher levels of subjectivity. With regard to human genetics, a challenge of reflexive modernity is to develop the means to conceptualise and evaluate the higher-level effects of these technologies.

Method and material

As the aim of the study was to search for ambivalence and reflexivity among individuals who in one way or another were committed to the utility of human genetics, we chose to interview these individuals in focus groups of 4-5 participants each. While the group setting provides a dynamics and a certain anonymity as compared with individual interviews, these small groups were more suitable for establishing the trust needed to discuss ambivalence than the conventional focus groups of 6-12 participants would have been. 13 scientists working with Decode were purposefully selected to achieve a fertile exchange of opinions and beliefs. To complement this material, we recruited four consecutive individuals among those volunteering blood samples and healthcare information to Decode.[15]

Focus group interviews lasting two hours were carried out by SH in four groups; two groups of 4 and one consisting of 5 scientists, the last one consisting of the 4 lay participants. A semi-structured approach was used, based on a short interview guide and basic principles for focus group interviews. The informants were asked to discuss any likely consequence of human genetic research in the lives of 'ordinary people'. They were encouraged to use examples from their own experience, and it was explained that the aim of the discussion was to explore rather than necessarily to agree on any definite views of the topic given. The interview guide included a strong commitment to take seriously any suggestions and points of view submitted by the

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informants, i.e. encouraging an earnest discussion of their merits, the relationship between different suggestions, etc.

The interviews with the scientists were conducted in English, while the lay participant interview was conducted in Icelandic. Full audiotape recordings were transcribed verbatim, with the recording from the lay participant interview being translated into English at the time of transcription. All transcripts were coded for emerging themes by SH and then analysed. The transcripts were subsequently coded and analysed by RS, and finally all three investigators agreed on a common interpretation after trying diverging interpretations against each other as well as against the theoretical framework, also seeking to take account of how the investigators' preconceptions might influence the analysis.[16]

Results

The results are presented under three headings, each section being introduced by typical quotes from the interviews.

1. Benefits, with epistemological, clinical and practical limitations

> *There is so much hope that this will be the key to everything. But I think we will never get there. You know, we are hoping that this could be a good diagnostic tool, that we will be able to develop drugs for everything that we will be able to diagnose. And then to correct diseases with gene therapy... If we're talking say 15-20 years we will know maybe a little bit more about things and … but I think to be able to cure things … it's far, far away from us. Even though we have all those tools it's going to be … But it can be helpful in some cases to be able to, like with pharmacogenomics to be able to tailor the drug therapy. ... I think the steps are all going to be very small. For example with the human genome project, when, you know, all the human DNA had been sequenced, people really hoped that it would, you know, open the door to almost everything. But it is a very small, minor step … that we just know the letters in there, but we don't know what they mean (scientist).*

Our informants – lay and scientists – all expressed the view that human genetics will bring great benefits to healthcare. The scientists were enthusiastic that their research was showing promising results, and they were confident that research will translate into healthcare benefits. The lay participants stated without hesitation that the research they were contributing to was important. This was the unmistakable 'baseline' point of view among our informants, and a major finding against which our subsequent results have to be evaluated. The reservations, concerns, second thoughts and ambivalence that we present below gain their relevance against this background.

Scientists and lay participants alike were unanimous that progress would be slow, as the way from basic genetic research to substantial clinical benefit is long and tortuous. The informants identified and discussed a catalogue of epistemological, clinical and practical limitations of genetic prediction and preventive measures derived from genetic information. Here, we will focus mainly on predictive testing.

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First, the epistemological value of predictive information derived from genetic testing will be limited, especially when it comes to testing for complex diseases and traits: Several scientists stated that it is "quite clear that predictions are uncertain" in the sense that even if an individual is found to carry a genetic disposition for a given disease it is far from certain that he or she is going to develop that disease. As an example of this, if Decode or others were to identify a genetic polymorphism correlated with alcohol abuse, there would be "a lot of people with this alcoholism-gene who are not alcoholics", according to one of the lay participants.

Secondly, it follows from the epistemological characteristics of predictive information that using this information as the basis of clinically relevant advice about lifestyle and preventive treatment is not straightforward. It is clearly a problem that diagnostic and prognostic technologies tend to be introduced ahead of evidence of their accuracy or the effectiveness of intervention. In addition it is unlikely that any preventive treatment will completely eliminate a given genetic risk. Also, a negative test result does not mean zero risk, and therefore leading a healthy life is important in any case. There will be uncertainty about potential genetic risks that have *not* been mapped, as stated by a scientist talking about a disease which runs in his family: "I would just be thinking 'is there another gene?'" The scientists agreed that making sense of genetic information and turning it into wise choices in everyday life would remain a hard task for most people, especially as information becomes more complex. When provided with information about multiple risk factors, people will have a hard time figuring out and adapting to the information: "[O]ne can wonder to what extent people will be able to understand these tests and to make use of them". The scientists also expected that the importance of genetic predispositions would be overestimated in some instances: "Some people might just go to bed and think, well my life is over, I have a three times risk of getting …" Some of the scientists wondered optimistically whether information technology could be used to 'translate' genetic information so that people could form appropriate opinions despite lacking the background knowledge necessary to evaluate the technical details of risk estimation and risk reduction. Then again, not everyone has the motivation to change lifestyle in accordance with medical evidence. Actually, the general knowledge that people should exercise, and avoid smoking and excessive consumption of food is available to everyone already, and still the lifestyle of the general public in most societies is becoming unhealthier by the minute. Some of the scientists wondered that this unfortunate trend might escalate as it becomes coupled with widespread use of lifestyle drugs designed to minimise the risk imposed by self-indulgence and inheritance. Unfortunately, the scientists said, many "people would rather 'pop a pill' than go to the gym". The misgivings of the scientists were largely corroborated by the lay participants. The avalanche of information from genetic research will be "an extremely large package" to people, as one of them put it.

2. Impact on dignity, tolerance and justice

> *When these individuals disappear from society, it will be: I'm precious ... my own needs. It will fail ... the sense of humanity. All this raises the need to sort out in general what is quality of life (lay participant).*

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The lay participants expressed a concern that genetic testing in pregnancy will increase the pressure to 'eliminate' abnormal foetuses. Although Decode has emphasised technologies targeting postnatal prediction and prevention of disease, the lay participants found it natural to include a discussion about reproductive technologies and suggested that dignity and humaneness are linked with diversity and tolerance in ways which make it dangerous for society to systematically 'eliminate' deviant individuals such as dwarfs and people with Downs syndrome.

The lay participants also wondered whether genetic research, although potentially useful, should be prioritised above other measures. When preventive measures based on genetic technologies are introduced, some people are going to demand them regardless of whether they are cost-effective in comparison with other interventions. The scientists discussed along the same lines that new technologies are demanding on resources, and genetic technologies may add to inequalities in health across social and global gradients. The possible creation of a 'genetic elite' was discussed, including references to popular culture (as in the film 'Gattaca') and popular science (as in the book 'Remaking Eden'[17]). All of this was seen as unjust and unfortunate.

3. Fighting disease and taking care of one's own health

> *I'm thinking about my [children]. How can I tune [their] life to avoid the stuff that are true risk factors today? Of course I want to understand this better. Give them proper food when they grow up, so they won't have the deficiency or whatever … signalling substance (scientist).*
>
> *And then it's the question ... er ... do you feel any better if you have this list from the moment you're born with all the syndromes that [starts to laugh quietly, the rest of the group gradually join] ... or, I don't know ... or that you might get? You're at high risk for this, and this and that. And if you are either going to live by this or oppose it and ignore it completely, what the heck, I'll just get everything. I don't know if that's a kind of knowledge that would actually be useful for one (lay participant).*
>
> *I think you have to balance the risk factors with what you can call the quality of life. You can worry yourself sick if you know too much (scientist).*

3A. Medical rationality and the empowering function of knowledge

The scientists sometimes argued as if medical rationality were the ultimate guide to a successful life. One should keep informed about one's dispositions to disease and make informed choices about lifestyle and preventive interventions. A scientist who until recently had been working as a MD, assured that this was how he used predictive information in his work, giving his patients "very concrete instructions about how they should behave and not behave in terms of various risk factors." The scientists said they hoped their research would lead to the discovery of genetic dispositions to disease that would allow people to take appropriate measures to avoid disease: "If we could identify those major players, then people could tailor their life." They emphasised the empowering capacity of predictive knowledge, i.e. that people can be put "in a more empowered role of being able to do something about it should they choose so."

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The general rule that one should follow medical advice was made more tangible when the scientists described how they would like to 'tailor' their own life or that of their children through use of medications, lifestyle-interventions and specialised diets, once "the true risk factors" become known. In one of the groups two female scientists also stated that if they were to learn that they were genetically predisposed to osteoporosis, they would respond by having their bone mineral density measured more frequently.

3B. Personal inconsistencies

In discussions about how the scientists themselves were currently dealing with issues of risk, it became evident that they are not always compliant with medical rationality as described above. For example, two of the scientists submitted that they were not having their cholesterol measured, even if they were positive that this would be prudent. Similarly, it was not always clear to the scientists that they would choose to avail themselves of preventive medications, though they expressed enthusiasm and support for research programmes designed to develop such therapies. For example, one of the scientists stated that she probably would not wish to take medications to prevent stroke, in spite of the fact that several of her older relatives have suffered from that disease: "I'm dying to know if I have that haplotype or not, but what I would do with it? I would not really change my life or anything. But would I take a pill to prevent ... based on that haplotype? No." In much the same way, the lay participant group was not willing to conclude that the teenage daughter of one of the participants would be better off knowing about whatever genetic dispositions she might be carrying.

The discursive devices employed by the scientists to explain apparent inconsistencies between the ideal of medical rationality and their own choices, included judgements about "stupidity and laziness", failure to make one's priorities correctly or inconvenience. These explanations were frequently coloured by laughter, signalling as well as containing unease about one's own shortcomings.

3C. The limits of medical rationality

On several occasions scientists and lay participants alike described potentially harmful effects of predictive knowledge and extensive preventive measures, and stated that prediction and prevention must not be made the only measure of how people lead their lives. This point of view is summarised in the following quote from one of the lay participants: "I think that people are going to ignore it, or I hope so [starts to laugh, quietly, the others gradually join the laughter]. That people don't live totally by … Because you have got to have a little freedom." The scientists were clear that no matter how sophisticated technologies that are developed, we will "never be free of the worries of being human with all its complications". Striving to eliminate every single worry will bring more misery than benefit. It did not become clear, however, under what circumstances this misery would outweigh the benefits of genetic technologies; and suggestions were made that people will gradually 'adapt' and learn how to make use of new information. In the following quote this is being discussed by a lay participant, but even here the use of laughter and the choice of Alzheimer's disease as an example seems to signal the unresolved nature of 'adapting' to predictive genetic knowledge:

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> *I think that one would just accommodate, just as everyone accommodates to knowing … well we know that eventually everyone is going to get some disease. It always takes time to adapt to what is new, but I think that one would just be glad to know that yes, I'm at risk for this. Then you'd be waiting for the first symptoms [starts to laugh] yes [laughs louder, the others join]. Then you'd know what it was: Now the Alzheimer is coming!*

Discussion

Although our informants demonstrated an ability to identify and discuss complex issues and concerns regarding the future of genetics, they may hold other beliefs on these topics than we have been able to elicit from these particular dialogues. Most obviously, the scientists may have had a strategic motive – perhaps encouraged by the interests of their employer – in portraying themselves as socially and morally responsible. Making a likeable and sympathetic impression is important to most people, and the scientists as well as the lay participants may have estimated that we – the investigators – as well as the future readership of this report would approve if they gave the impression that they were concerned about potential harm caused by genetics. As stated above, however, we have approached our informants with a strong commitment to take seriously their arguments and stances, and in the following we will pursue the lines of thought communicated in the interviews without further speculation about vested interests.

Taking Wynne's idea about the prescriptive content of science and technology as our point of departure, the question to be answered is what we have learnt about how our informants relate to the prescriptive messages of human genetics. As indicated above, the organising principle of our results is that this relationship is characterised by an ambivalent mixture of enthusiasm, second thoughts and reservations. We will now discuss certain features of this mixture.

Our informants' general enthusiasm about the benefits of human genetics and the scientists' ideas about personal lifestyle choices and preventive treatment presented in section **3A** illustrate a particular prescriptive model of human agency and the measures that should be taken to avoid disease. According to this model individuals are endowed with the capacity to make rational decisions based on objective knowledge about what is to their advantage or disadvantage. Based on predictive knowledge about their predispositions to disease, individuals are obliged by reason to make such decisions as minimise the likelihood that they fall ill. Each person's life should be 'tailored' with the aim of securing his or her health, and this is done by making the right lifestyle choices and benefiting from preventive medications in order to minimise specific disease risks. This prescriptive model, which we will label 'rational lifestyle choice' has strong foothold in research and healthcare policy in Western societies, and is part of the cultural milieu hosting and encouraging the rise of human genetics.

Already the acknowledgement of the limitations of predictive knowledge presented in section **1** of the results betrays that our informants are aware that this prescriptive model has serious limitations. Presupposing that people will make rational lifestyle choices based on information about a multitude of risks, that they enjoy the capacities

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needed to make all the 'right' choices, etc., runs counter to the common knowledge that awareness of lifestyle influences on health does not in itself guarantee that people 'tailor' their lives towards 'minimal risk'. In general, the very belief that there is a 'right' way of using genetic information, and the assumption that people *can* and *will* use genetic information in the 'right' way, are problematic. This emerges from the probabilistic nature of genetic information[18] and the effects genetic technologies may have on the non-biological dimensions of health discussed by van Hooft. While genetic testing and preventive regimes increasingly provide opportunities for countering biological disease, the crucial insight is that these regimes *also* have the potential to interfere with the autonomous self-expression and spontaneity of individuals and societies which are essential ingredients of health in the non-biological sense described by van Hooft. Deciding which are the 'right' conclusions concerning the use of preventive regimes based on genetic technologies is therefore likely to be a much more complex task than suggested by the prescriptive model of rational lifestyle choice.

The results presented in section **2** demonstrate our informants' attempts to explore how the prescriptive model implied in their most enthusiastic interpretation of genetics may impact other values and prescriptive beliefs, in particular the social virtues of dignity, tolerance and justice. Even if human genetics will increasingly improve biological health for privileged individuals, there is a danger of reduced tolerance of diversity and more generally a threat to the sense of meaning in people's lives, placing human dignity and sociality at peril. Under certain conditions the poor and underprivileged are likely to suffer increased injustice and deprivation in comparison with people enjoying good access to new genetic technologies. Although our informants did not dismiss genetic research on these grounds, or even conclude that human genetics is bound to cause intolerance and injustice, their discussion nevertheless illustrates that these adverse effects may occur, particularly if enthusiasm for human genetics is unrestricted and based on a particular set of prescriptive beliefs, valuing individual 'tailoring', risk reduction and enhancement to conform with narrowly conceived concepts of health and success. Thus, our results indicate that both lay people and scientists strongly in favour of genetic research and technology development, see the prescriptions of a narrow medical rationality as inadequate if genetic technologies are to be compatible with human dignity, tolerance and justice.

In sections **3B** and **3C** second thoughts and ambivalence are brought closer to our informants and their personal points of view. Some of the scientists betrayed that they were not personally following the lifestyle obligations to which they were otherwise eager to claim support. And neither the scientists nor the lay participants were certain that they would change their lifestyle or take medications in accordance with predictive knowledge based on genetic tests in the future. Within the rational choice model this amounts to nothing less than foolishness, and it is therefore understandable that our informants reacted by making fun of themselves and each others on these occasions. Especially for the scientists, any apparent failure to adhere to 'scientific standards' can easily become a cause of embarrassment. Acknowledging and underscoring one's humanness by use of laughter and similar devices can be a way of glossing over or implicitly explaining one's failure in everyday life to adhere to a scientific worldview, of which the rational choice model is an integral part.

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The points of view presented in section **3C** constitute the most explicit reservations in the face of the prescriptive model of rational lifestyle choice found in our interviews. Here our informants, who through their enthusiastic work or through unselfish donations are committed to the idea that human genetics can provide relief and improve health, are explicit in their reservations against a lifestyle where prevention and 'tailoring' are the supreme measure of how one should lead one's life. Interestingly, ambivalence and reservations in the face of enthusiasm appear to be inherent features of their positions, rather than a temporary state of doubt to be resolved by an argument either in favour or in disfavour of human genetics.

Taking our lead from the statements "you have got to have a little freedom" and "you can worry yourself sick if you know too much", we believe that the ambivalent stance demonstrated by our informants can be explained in the light of van Hooft's theory of health. Human subjectivity is constituted not only by biology, but by the ways the organism relates to its environment, constructs meaning, partakes in social relationships and commits itself to certain ways of living, and genetic technologies designed to prevent or combat organic disease, can interfere with health on all these different levels. Thus, *our informants' ambivalence can be understood as the logical consequence of reacting to genetics on two or more health levels at the same time*: They are enthusiastic that genetic technologies can be useful to avert disease, but simultaneously suspicious that extensive predicting and 'tailoring' can have unintended and unfortunate effects on the creative processes involved in maintaining relatedness, purpose and meaning. Further, scientists and lay participants alike conducted their non-conclusive negotiations between contradictory opinions and sentiments in a quite identical manner, indicating perhaps that the basic requirements for assigning a meaningful and beneficial lifeworld role to medical technologies are shared by all subjects, and are not strongly influenced by technical expertise in the field in question. As reported in section **3A** the prescriptive model of rational lifestyle choice emerged most clearly from the dialogue among the scientists, but their commitment to this model was balanced by equally strong statements of reservation, ambivalence and non-adherence indistinguishable from those of the lay participants.

As genetics is currently held to be among the most promising venues of research to further enhance the power of medical interventions against disease, pain and disability, any second thoughts about human genetics can be seen as spitting in the face of medicine's technical miracles. Further, medicine is thoroughly embedded in a scientific parlance of objectivity and disenchantment. Non-biological levels of health on the other hand, being intrinsically dependent on cultural and subjective interpretation, do hardly lend themselves to rigorous analysis and experimentation. Suggestions that genetics could be harmful to health – to meaning, agency, belonging, etc. – can therefore easily be dismissed as unscientific speculation, not suitable for rational validation or refutation, and therefore sheer nonsense. The very idea that regimes which effectively combat disease can be harmful to health is probably counter-intuitive to many. Our suggestion is that this results from the biological one-dimensionality of Western medical and scientific epistemology: By neglect, this epistemology and the corresponding prescriptive beliefs currently in circulation do not distinguish, hence do not weigh benefits (or losses) on the biological dimension of disease against losses (or benefits) on non-biological levels of health. Implicitly, health is treated as a non-existent dimension in life; or rather the concept of health is

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reduced to imply merely the absence of disease, or the 'normal' functioning of organs.[19]

Conventional risk communication has been shown to be at best an imperfect method for recruiting the general public to the regimes of preventive medicine. This has given rise to the understanding that individuals' and populations' health needs, values and aspirations must be explored as they provide the context within which the merits of medical technologies are to be established. Thus, it is an implication of our study that the non-biological dimensions of health should be taken into account when judging the proper role to be assigned to human genetic technologies.[20]

While our informants discussed the material costs of new technologies and the intellectual challenges involved in interpreting estimates of disease susceptibility, it seems to us that cultural resources in a broader sense may be equally important for people's ability to make sense of genetics. The decisions people make regarding their health are a product of their knowledge, imagination and sensitivity, vulnerable to anxiety, lack of insight and social support. Information and choices that may empower the privileged who have access to cultural resources to sort out their options, may be misinterpreted by others, causing suffering, loss of sense of good life, etc. Thus, one of the reasons why health is strongly related to class and social status[21] appears to be that people need freedom, knowledge and social support to take good care of their health and to benefit from healthcare. We submit that this hinges on, among other things, people's ability to balance any unintended and unfavourable effects of medical technologies on the non-biological dimensions of health against their intended and favourable effect against biological disease.

In the face of van Hooft's model of health, it should be stressed that there is no way of knowing *a priori* the size or the sign of effects of genetic technologies on non-biological levels of health. Indeed, returning to Tomas' and Thomas' theorem, it becomes clear that the public reception of genetic technologies in itself is medically relevant. And if the higher-level effects of new technologies are dependent on the cultural interpretation of the same technologies, the task of exploring and evaluating the higher-level effects of human genetics takes a creative turn: In addition to its analytical part, we need to mobilise the cultural and governance resources which will ensure the compatibility of genetic technologies with the full range of human subjectivity and health. This might produce some rather startling effects. Perhaps the private discourses of ambivalence and reflexivity may be regarded as a resource for the development of the required adaptation: their significance in this respect needs clarification in further research.

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[1] See for example: European Comission. 2003. *Europeans and Biotechnology in 2002*, Department of health. 2003. "Our inheritance, our future, realising the potential of genetics in the NHS", available at: http://www.doh.gov.uk/genetics/whitepaper.htm., and Wellcome Trust. 1998. *Public perspectives on human cloning.* London.

[2] T. M. Bubela and T. A. Caulfield. Do the print media "hype" genetic research? A comparison of newspaper stories and peer-reviewed research papers. *Cmaj.* 2004;170(9):1399-1407.

[3] A. Petersen. Biofantasies: genetics and medicine in the print news media. *Social Science & Medicine.* 2001;52(8):1255-1268.

[4] Cf. the project 'Public understanding of genetics': http://les1.man.ac.uk/sa/pug/index.htm. and B. Wynne. Creating public alientation: Expert cultures of risk and ethics on GMOs. *Science as Culture.* 2001;10(4):445-481.

[5] A classical account of reflexive modernity and the scientific construction/production of risk is found in: U. Beck. *Risk society: towards a new modernity*. London. Sage: 1992, while the following papers demonstrate how these ideas can be applied to the field of genetics and health: A. Kerr and S. Cunningham-Burley. On Ambivalence and Risk: Reflexive Modernity and the New Human Genetics. *Sociology*. 2000;34(2):283-304, and E. Beck-Gernsheim, Life as a planning project. In: S. Lash, B. Szerszynski and B. Wynne, eds. *Risk, environment and modernity.* London. Sage publications: 1996.

[6] Cf. L. Frewer. Risk perception, social trust, and public participation in strategic decision making: Implications for emerging technologies. *Ambio.* 1999;28(6):569-574, and J. G. Roederer. Communicating with the public, politicians and the media. *COSTED Occasional Paper*. 1998;NO 1:1-28.

[7] A. Árnason and B. Simpson. Refractions through Culture: The New Genomics in Iceland. *Ethnos.* 2003;68(4):533-553. For balanced accounts of the controverses raised by Decode genetics see for example: G. Pálsson and K. E. Harðardóttir. For whom the cell tolls: Debates about biomedicine. *Current anthropology.* 2002;43(2):271-287, and J. F. Merz, G. E. McGee and P. Sankar. "Iceland Inc."? On the ethics of commercial population genomics. *Soc Sci Med.* 2004;58:1201-1209.

[8] European Comission. Special Eurobarometer 225: Social values, science & technology. Available at: http://europa.eu.int/comm/public_opinion/archives/ebs/ebs_225_report_en.pdf.

[9] K. Kristjánsson, personal communication, May 12th, 2004

[10] Decoding the language of life - A case study of deCODE genetics in Iceland. Available at: http://www.uib.no/isf/people/stefan.htm.

[11] B. Wynne. May the sheep safely graze? A reflexive view of the expert-lay knowledge divide. In: Lash, Szerszynski and Wynne, *op. cit.* note 5.

[12] B. Wynne. Creating public alientation: Expert cultures of risk and ethics on GMOs. *Science as Culture.* 2001;10(4):445-481.

[13] W. I. Thomas and D. S. Thomas. *The Child in America: Behavior problems and programs*. New York. Knopf: 1298.

[14] S. van Hooft. Health and subjectivity. *Health.* 1997;1(1):23-36, and S. van Hooft. Disease and subjectivity. In: J. M. Humber and R. F. Almeder, eds. *What is disease?* Totowa, NJ. Humana Press: 1997.

[15] Among the 13 scientists, there were 6 women and 7 men, 8 were Icelanders and 5 non-Icelanders, 4 were doctors and 9 biologists. The group of lay participants comprised 3 women and 1 man; the man stating that he had been recruited for Decode's research because he was affected by a certain disease; one of the women stated that she was related to someone suffering from a disease being researched; while the remaining two did not volunteer information about the reason for their recruitment for Decode's research. The inclusion of the lay participant group was approved by the Icelandic Research Ethics Committee, and their anonymity was secured by use of pseudonyms and exclusion from the transcript of any information from which their identity could be inferred.

[16] The general methodology applied is roughly equivalent to that described in R. Barbour and J. Kitzinger, eds. *Developing focus group research*. London. Sage Publications: 1999. For a closer description of the analytical strategy employed, see: R. B. Addison. A grounded hermeneutic editing approach. In: B. F. Crabtree and W. L. Miller, eds. *Doing qualitative research*. Thousand Oaks. Sage Publications: 1999, while the following paper by K. Malterud details some of the requirements for reflexivity on behalf of the investigator: Qualitative research: standards, challenges, and guidelines. *Lancet.* 2001;358(9280):483-488.

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[17] L. M. Silver. *Remaking Eden: How genetic engineering and cloning will transform the American family*. New York: Avon Books; 1998.

[18] While they were not asked to discuss genetic exceptionalism, our informants consistently maintained that genetic technologies being developed today will provide probabilistic prediction and prevention of disease in the same way as do conventional medical technologies. On this issue at least they agree with the current understanding that genetic information shares crucial features with other medical information. For a recent discussion of genetic exceptionalism in the context of common diseases, cf. the European commission's Expert group on genetic testing report *Ethical, legal and social aspects of genetic testing: research, development and clinical applications.* Brussels, 2004.

[19] Humber and Almeder, *op. cit.* note 14.

[20] Ideally, the increasingly applied public health methods of Health Needs Assessment and Health Technology Assessment take into account cultural, social and ethical aspects of their subject matter, which we take to include acknowledgement that although immensely valuable to individuals and societies, the treatment and prevention of biological disease must also be evaluated in terms of its impact on other values, including the non-biological levels of health. The UK National Institute for Health and Clinical Excellence (NICE) provides an illustrating example of systematic and prospective assessment of the technical and public value of medical technologies, cf. especially http://www.publichealth.nice.org.uk/page.aspx?o=513203, accessed on December 14th 2005.

[21] Cf. R. G. Evans, T. R. Marmor and M. L. Barer, eds. *Why are some people healthy and others not?* New York. Aldine de Gruyter: 1994, and R. G. Wilkinson. *Mind the gap: hierarchies, health and human evolution*. New Haven. Yale University Press: 2001.

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Africa must come on board the genomics bandwagon

TEMIDAYO O. OGUNDIRAN

Abstract

With the completion and the success of the unraveling of the human and some non-human genetic codes comes the optimism that science, once again, is at the threshold of transforming human existence in an unprecedented way. The sequencing of the human genome with the science and technology by which it occurs is seen as a potential gateway to man's final conquest of most of the health and health related disorders that have for long plagued the human race.

While some developing nations like Cuba, Mexico and India have taken the initiative to be major players in the genomics arena by exploring its potentials towards enhancing improved quality of life for their citizens, the majority of others, especially in Africa, still occupy the spectators' seat. If this apparent lukewarm attitude continues, it implies that the present dependence on the developed nations as remote beneficiaries of gains of scientific breakthroughs will persist. It also means that the expectation that genomics should, among other benefits, re-dress the inequalities of access to health care between the rich and poor nations may be a mirage for a long time to come.

The expected benefits from the human genome project and genomics technologies are fascinating and hold the ace for improving the standards of living of the African people. However, these expectations are futuristic, time-dependent and capital intensive. They require commitments of national governments to policy re-orientation about research and development, strategic planning, resource mobilization, priority setting; and establishing, promoting and sustaining enabling environments for scientific and technological breakthrough. No doubt, there is dire need for assistance from the developed and the frontline developing nations in this regard. However, great initiatives and deep commitments to making significant scholarly contributions to the advancement of biotechnology and its potentials in the near future must come from within.

Background

The Human Genome Project, straddling the close of the 20th and the beginning of the 21st centuries, is at the threshold of changing human thinking both in the way we look at the past and in the way we view the future. Genomics has the potential to unravel the mystery of past existence, modify our understanding of present events and re-direct our focus on things that enhance the future good of humanity. Though its applications and uses appear to be more direct to medicine and human health, it has the potential to transform human civilization beyond the confines of biomedicine. The discovery of antibiotics in the early part of the last century marked the beginning of the march towards the conquest of infectious diseases resulting in total eradication of some and complete control of others, especially in the developed world. Genomics is to this century what infectious disease and discovery of antibiotics were to the last.[1]

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The potentials of genomics, however, transcend those of antibiotics in that it holds promises to revolutionize our knowledge about the management and control of both infectious and non-infectious diseases.

Of particular importance is the relevance of genomics and its tools to narrowing the health gap between rich and poor nations and enhancing reasonable access to simple, cheap and effective health services in developing countries.[2] In view of this potential, calls have been made to less developed countries of the world not to stand by in the mistaken impression that genomics is of and for the developed world. Some developing nations like Cuba, Mexico and India have risen up to the challenge and have taken the initiative to be major players in the genomics arena by exploring its potentials towards enhancing improved quality of life for their citizens.[3] However, many countries of Africa excluding South Africa, Egypt and perhaps one or two others, still lag behind. It is imperative that African nations emulate the examples of the developing countries that have assumed frontline positions in genomics.

In a foresight study of the ways in which genomics is likely to affect the third world, Daar et al identify top ten technologies for improving health in developing countries.[4] Prominent on the list is the use of genomics biotechnology to confront the menace of infectious diseases through development of simple and affordable diagnostic methods; development of vaccines and efficient delivery systems; and identification of new antimicrobials and drug targets. The list also includes application of genomics to improving agricultural yields and enhancing a clean environment. Pang and many others support the view that genomics would enhance the control of infectious and non-infectious diseases on a global scale. Further more, sequencing of the rice genome offers opportunities for improved yield and more nutritious value for the three billion people, mainly in the developing world, who depend on rice as their staple diet.[5] [6] These and many others express the common view that genomics could be a veritable tool for improving health and living standards in developing countries. This paper focuses on developing African nations using Nigeria with immense potentials as a prototype, not only to contribute to the genomics breakthroughs, but also to reap from its windfall. It explores why African nations should not lag behind other developing nations of the world in exploring the genomics landscape and considers the process towards achieving this drive.

Discussion

The situation: Nigeria as a prototype

With a population of 130 million people, an annual growth rate of about 2.8%, more than 250 ethnic groups and over 300 dialects and languages, Nigeria is the most populous and most culturally diverse black nation in the world. One in two West Africans and one among six Africans is a Nigerian. The country emerged from the ruins of a civil war in the late sixties and early seventies to a dawn of a new era signaled by the discovery of abundant petroleum oil reserve in the Niger delta. This discovery resulted in total abandonment of the agriculture sector which had hitherto been the mainstay of the economy, and an over-dependence on the capital-intensive oil sector, which now provides almost all of foreign exchange earnings. What began as an “oil boom” with a lot of potentials for development and a strong foreign

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exchange earning capacity and economic base for the country later became an albatross which may now be looked back upon as an "oil doom". A combination of failure to further develop other sectors of the economy, over-dependence on petro-dollar, military mis-adventure in power with consequent destruction of the moral fabrics of the society and an enthronement of systemic corruption, mismanagement, inflation, political unrest and ethno-religious conflicts, has produced a state with a GDP per capita income of about US $300.

However, except for a few setbacks which are reminiscent of a long period of military rule, the return of participatory democracy in Nigeria in the past six years has brought about significant re-orientation of the body polity. The relatively stable political climate should provide a springboard for an all round development and an incentive for investment in science and technology. Though the potential gains of genomics technology are still a long way to reach even in the technologically advanced countries of the west, it is imperative for resource- poor and less technologically advanced nations to join forces with others now. They must not wait till the gains become tangible, otherwise those benefits would be out of their reach and the present status quo of disparity in distribution of the gains of science and technology in health would remain, and even worsen. Nigeria and the whole of Africa stand to benefit in many aspects especially in health, agriculture and the environment.

Health profile

Infant and child mortality rates are basic indicators of the socioeconomic development of a country and the quality of life of its people. Specifically, infant mortality (which is the probability of dying before the first birthday) in Nigeria is highest when compared with some countries from different geographic zones of the world (Figure 1). In the period from 2000 to 2005, while the infant mortality rates in most of these countries (except South Africa and Iran) have fallen consistently, the rate increased from 74.18 to 98.8 per 1000 live births in the same time period in Nigeria.[7] The high prevalence of communicable but preventable diseases like malaria, dysentery, pneumonia and measles contributes significantly to high childhood mortality and morbidity rates in Nigeria. Besides these are other infectious diseases like typhoid, amoebiasis, guinea worm, helminthiasis, tuberculosis and hepatitis which also cause considerable mortality and morbidity in both adults and children. While most of these communicable diseases have been eradicated or controlled in the developed and some developing countries, they still constitute a major health hazard in Nigeria and most countries in sub-Saharan Africa.

Moreover, a significant proportion of Nigerians have genetic disorders such as sickle cell anaemia and glucose 6-phosphate dehydrogenase deficiency or their traits. In addition to the burden posed by infectious and genetic diseases is the epidemiological transition which Nigeria, like many other developing countries is undergoing.[5] Hypertension and diabetes presently affect about 10% and 2.75% of the population respectively, but prevalence rates of these diseases are rising.[8] The incidence of breast cancer appears to have stabilized and is perhaps declining in some western countries, whereas it has been increasing in those societies that hitherto had a low incidence including Nigeria.[9] Regrettably, little or no attention is given to these time-dependent fatal diseases some of which are hinged on lifestyle and behaviour.

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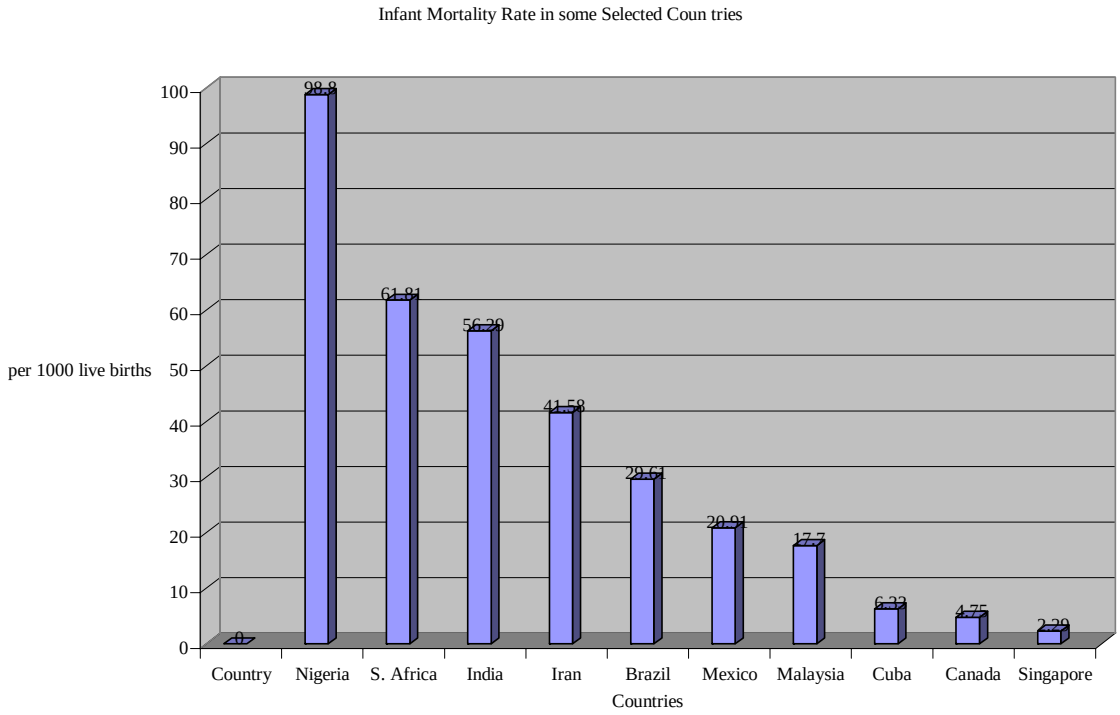
Figure 1



How genomics (proteomics, pharmacogenomics etc) might help the situation

One of the promising benefits of the knowledge of human and non-human animal genomes is the application of technology to enhance the health of humans in both the developed and the developing worlds. As is shown above, the major causes of morbidity and mortality in Nigeria and most African countries are preventable, treatable and controllable. Most of these diseases present a formidable prevention, diagnosis and treatment triad. Apart from an efficient public health system and a positive attention to personal and environmental hygiene by the people, the only other significant means of curtailing the spread of these communicable diseases is an effective vaccination programme. Hitherto the different vaccination programmes and efforts in Nigeria for example, are far from being universally successful, adequate or effective. In spite of the fact that most of the vaccines are supplied free by donor nations and agencies, and are provided free to the public, some other extenuating factors like difficulty with maintaining an unbroken cold chain, non-oral route of administration and cumbersome dosing schedules militate against effective vaccination against the killer diseases. It is hoped that advancements in genomics and DNA based vaccine production would result in developing vaccines whose availability, storage and administration will bypass or overcome most of the hurdles hitherto encountered in immunization programmes. The prospect of utilization of plant science as a new vehicle for vaccine delivery holds good promise for Nigeria and the whole of Africa.[10 11]

Vector control through genetic modification and biochemical targets offers a promising approach in the battle for conquest of many diseases. At the Africa Human Genome Initiative Conference in South Africa in March 2003, Gordon Dougan,

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Director, Centre of Molecular Microbiology and Infection, Imperial College, United Kingdom, told participants who came from African countries: “Don't be hypnotised by technology, vaccines can be produced with relatively simple technology within five years..”[12] Talking about developing vaccine against typhoid infection, he said: “There is no great mind boggling technology involved in this - it is relatively simple, so don't be scared of it.”[12]

Another major health hurdle is the diagnostic difficulties that clinicians and laboratory scientists encounter which make pathogen isolation an arduous task and which invariably delay institution of definitive drug therapy. The new biotechnology would facilitate development of simple, rapid and high precision diagnostic tools which would be available and applicable in the developing countries where they are most needed and where there is a ready market for them. Presently most new drug research and development are done in western nations and of all the 1233 drugs in the market between 1975 and 1999, only 13 were for diseases that are prevalent in the tropical countries.[13] Through pharmacogenomics and with the increasing knowledge of the genetics of diseases and the genetic variations among groups of peoples, it should be possible to develop new drugs against many tropical diseases. Such drugs would be selective, efficacious, safe and better tolerated. Moreover, in a continent where trauma and thermal burns with large wounds from vehicular and fire accidents are rampant, biotechnology holds a promise for developing living cultured skin for managing the wounds.[14] [15]

Agricultural profile

Farming remains a major income-generating occupation of many urban and rural dwellers in Africa. Many people engage in farming as a means of supplementing their meager income or providing food for their households and extended family members; while some others practice farming as a commitment to a long family tradition. Apart from having a favourable agro-climatic condition, it is estimated that Nigeria has a cultivable land area of 71.2 million hectares and an agricultural population of about 38 million people. This agricultural population is said to be 3 times the combined agricultural populations of Japan, USA and the UK.[16] [17] Arguably more than 50% of Africa’s active labour force is employed in agriculture. As alluded to above, Nigeria had been a world class producer of cash crops such as cocoa, palm produce, rubber and groundnut before the oil boom era of the 1970s.

In spite of this potential of becoming an agricultural giant in Africa, Danladi Kuta, a molecular biologist with the Sheda Science and Technology Complex, Abuja, Nigeria says Nigeria remains an agricultural dwarf and a low-income food-deficit country.[17] Nigeria does not produce enough to feed its teeming population and imported food products worth millions of dollar in the last decade. Many wage earners spend most of their income on food alone. According to him, the current agricultural growth rate of 4.5% is far less than the 10% mark which is the minimum that is required to meet the increasing food demand of the nation. The reasons for the country’s low agricultural productivity include the fact that the bulk of agricultural production is done by resource-poor rural farmers who plant crop varieties with poor yield and that can not tolerate water stress or resist insect pests and infestation with nematodes.[17] Other factors include poor weed control strategies, post-harvest storage constraints and largely a non-mechanized farming system.

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How genetic modification (GM) technology might help the situation

Jacques Diouf, Food And Organization Director-General, states that "biotechnology offers opportunities to increase the availability and variety of food, increasing overall agricultural productivity while reducing seasonal variations in food supplies." He states further that "the effective transfer of existing technologies to poor rural communities and the development of new and safe biotechnologies can greatly enhance the prospects for sustainably improving agricultural productivity today and in the future"[18] Apart from the developing countries of Africa, even in the developed countries of the world, there is increasing demand for more and better quality food as the world population increases and the quantity and quality of arable land mass diminish as a consequence of overcrowding, industrialization, land degradation by over tillage and organic pollutants; all leading to "an increasingly fragile natural resource base" that will be incapable of providing enough food for the growing world population.[19]

Though there is a global need for improved food quality, the situation becomes more precarious in Africa where most under-five deaths are associated with malnutrition. I agree with Kuta that an investment in agricultural biotechnology would lead to a mitigation of the constraints that presently hinder bumper yields through development of herbicide-tolerant transgenic crops, improved crop varieties that can tolerate water stress and crops developed with resistance genes to insect pests, nematodes and viruses. According to him, glyphosate is a safe and non-residual herbicide, but it is non-selective and destroys green plants; hence it has to be sprayed before or just after planting the crops thus limiting weed control to the initial phase of cropping. However, through GM technology, glyphosate-tolerant soybeans and corn are now grown in some developed countries. African farmers would benefit from the application of similar technology to locally produced crops like rice, beans and guinea corn which are major food crops across the continent. Cowpea, cassava and yam are major staple food sources, which are plagued by insect pests, viruses and nematodes. Incorporation of pest resistance genes into their genomes is one way of improving their survival and enhancing good yield. Further more, development of cyanogen-free transgenic cassava and incorporation of gluten genes into cassava will lead to improved cassava quality as well as cutting the present foreign exchange spending since cassava flour could be substituted for imported flour used in the baking industry.[17]

Environment and bioremediation

The principle of bioremediation has wide applications in developing countries as a cheap and effective method for cleaning up the environment through the use of natural and genetically engineered microorganisms.[4] This technology is of particular benefit to Nigeria where sewage disposal, water pollution and petro-chemical oil spillage with consequent destruction of natural plant and marine life especially in the Niger Delta are major health hazards. In Brazil, the Federal University of Santa Catarina and Petrobrás, the Brazilian national oil company jointly address the degradation of benzene, toluene, ethylbenzene and xylene (BTEX) from gasoline spills by phytoremediation, a bioremediation process that uses plants.[20] By investing in biotechnology, the Nigerian government would be providing the initiative for the

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several multi-national oil companies operating in the Niger Delta to team up with local universities in tackling the huge problem of environmental pollution through bioengineering.

The process

Change of mindset

The initiatives for change have to come from within Africa and not only from without. The hitherto lukewarm attitude of African governments to research and development has to give way to informed commitment by governments and all stakeholders. Proactive steps and critical policy shifts should move the continent from being a recipient of technology end-products (from the west) and from being a predominantly consumer economy to a technologically productive continent. If the developed and the up-coming nations of the world have achieved significant momentum in biotechnology and are tapping its benefits through concerted efforts of both their governments and peoples, the process cannot be otherwise for Africa.

The process should begin by learning from the examples of frontline developing countries like Mexico and India, by enacting policies that are backed up with legislative power, and by commitment to building a robust infrastructure that is immune to political control and manipulation. Governments must provide conducive environments, tax incentives, and favourable land lease agreements for local and foreign investors that show interest in investing in biotechnology. As is the case in South Korea, patent protection incentives should be given to scientists and entrepreneurs who venture to invest in biotechnology.[21]

Comprehensive education program

There is a long phase between scientific and technological breakthroughs and their final consumable end products. In this regard a well informed and motivated citizenry is a great asset. Public understanding is very important especially with regards to information about the eventual end products of genomics and biotechnology, for example, the cultural acceptability of GM food. Adequate education programmes are needed at all levels to enhance public understanding of the concepts of biotechnology, genomics, genetic engineering, genetic testing, genetic research, genetically modified food etc. This can be achieved through appropriate utilization of different educational programmes, schools, public fora, the news media, NGOs etc. Needless to say that molecular biology and genetics should be an integral part of the training of physicians, nurses, other health care professionals and biochemical scientists.

Building and strengthening capacity for biotechnology

The success or failure of a scientific innovation or endeavour is critically dependent on many factors including the comprehensiveness of the capacity that is acquired prior to inception of the project and the mechanism that is put in place for lubricating and strengthening the system for on-going efficient and satisfactory performance. Presently, pharmaceutical and related industries in Nigeria and most African countries do not possess the biotechnological capabilities of the developed world. For example,

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none of the national crop research institutes in Nigeria has the appropriate facilities and the critical mass of scientists for the application of GM technology except at the International Institute of Tropical Agriculture (IITA) which is presently developing transgenic banana/plantain, cowpea, and cassava.[17] Therefore, establishing a solid biotechnology base for Africa requires equipping academic centres and research institutes with infrastructure for molecular science and technology. Human capacity building would be enhanced by establishing scholarships for undergraduates who are willing to pursue a career in molecular sciences and by providing funding for graduate students to do research in molecular biology, biotechnology, bioinformatics etc. Governments should be committed to increasing spending on research and development, increasing education budgets at all levels, and prioritizing the teaching and learning of mathematics, the sciences and technology as was the case in India immediately after independence in 1947,[22] and as is presently done in South Africa. Moreover, other African nations should emulate the example of South Africa in developing and articulating their biotechnology sector to address their specific health needs.[23]

African governments should establish a network of researchers within their countries, coordinated regionally or nationally, and launch specific biotechnology development targets periodically. Moreover, governments should court the good will of frontline developing countries that have made significant progress in the field and seek to sponsor academics to fellowship programmes in such countries and also establish exchange of researchers with them. Academic institutions should be encouraged to pursue collaborations between local and foreign scientists both from developed and developing countries. Governments should also midwife industry-institution partnerships between the universities or research institutes and pharmaceutical and allied industries.

Fostering regional collaborations within African nations

On 3rd May 2004, the Nigerian National Biotechnology Development Agency in Abuja, Nigeria, launched the Nigerian Agriculture Biotechnology Project (NABP) and the West African Biotechnology Network (WABNET).[24 25] According to the Nigerian Minister of Science and Technology, the project would help to raise the effectiveness and efficiency of bio-resources and biotechnology in Nigeria and West Africa; and promote economic growth, ensure sustainable use of the natural resource base, enhance the health, environmental, industrial and agricultural development in the sub-region. This kind of collaboration between Nigeria and its neighbours is akin to the Asia-Pacific International Molecular Biology Network (IMBN) and European Molecular Biology Organization (EMBO). More of such collaboration is needed in Africa in molecular biosciences and biotechnology. It encourages cross fertilization of ideas and presents a common front for tackling health and health-related problems that are peculiar to the sub-region and continent. Similarity of circumstances would make these programmes more relevant and cost effective.[25] Given the strategic position, the human and economic potentials of Nigeria in the West African sub-region, this could begin a process of diversifying its economy by exporting biotechnological assistance to neighbouring countries that would in turn obtain it cheaper than they would from developed nations.

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Resource mobilization

The foreseeable gains of genomics are not reliant on biotechnology alone, but also on politics and resources.[26] Resource mobilization for biotechnology involves a re-prioritization of national interests and programmes and a re-distribution of the available resources to high priority and promising areas. Nigeria runs a wasteful political system that spends much resource to service and maintain the different arms and functionaries of government. The civil service is too unwieldy and unproductive to justify the huge wage bill that the different tiers of government incur every month. It is time Nigerians and other Africans realized that politics and civil service are no longer avenues for sharing the so called "national cake", and that remuneration should be based on productivity and relevance.

Justice requires that priority funding is given to programmes that benefit the continent the most. Traditionally, a large percentage of budgetary allocation goes to many unproductive services, an example of which is defense. The defense budget in Nigeria and perhaps in many African countries supports a retinue of highly redundant military personnel some of whom are highly qualified but underutilized in productive service to the country. No doubt, the Nigerian military consists of a crop of young, brilliant and science-oriented officers that could be linked up with academic institutions in the country with a mandate to produce a biotechnology revolution for the country. In addition, it is time rich and affluent Africans were re-orientated to embrace the culture of supporting science and health related research. Philanthropic investments and endowment of chairs in biotechnology in academic institutions by well-placed and affluent Africans might be a better way of investing in the future of the continent. Besides all these, apart from the initial lump sum as take-off grants, governments should commit sizeable percentages of their annual budget allocations to the biotechnology industry.

Enforcement of the rule of law

The bane of most developing countries of the world is political instability and corrupt leadership. A stable polity and an accountable and responsive government are the minimum requirements for development and for attracting foreign investors. The present political stability in Nigeria, Ghana and many other countries of Africa coupled with the active involvement of their present leadership in the New Partnership for African Development (NEPAD), established to support economic and political reforms in Africa through peer review of one another's performance, is a positive development for the continent. What needs to be put in place is due ethical and legal process for establishing and enforcing appropriate regulatory standards for genomics research and mechanisms for ensuring financial prudence and fiscal discipline. Furthermore, both the public and private sectors should collaborate in common biotechnology ventures. The private profit-oriented partner provides the fiscal and entrepreneurial discipline and thoroughness that are often lacking in government run programmes, while the public component regulates and controls the profit driven private sector interests. Also important to consider under legal considerations are intellectual property rights and patent issues. Developed countries will be reluctant to share technologies with developing countries legal systems that do not entrench and enforce intellectual property rights. In many African countries, intellectual property rights are either non-existent or are grossly violated and so patent

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holders are reluctant to share technologies with these countries. This is a major impediment to realizing technology breakthrough in Africa and African governments and legal systems must address it forthwith.

Ethical issues

Genomics and biotechnology raise many complex ethical, legal, and social concerns and challenges which require that national bioethics policy be instituted to guide the development of the biotech industry and protect the interest of the people. International safety and ethical standards should be adapted in the context of local factors such as literacy level, socio-economic conditions, cultural practices and religious beliefs. Capacity building in bioethics should be pursued pari passu with the evolution of genomics science and technology by providing training for physicians, nurses, other health care professionals and biological scientists. Special training is required for research ethics review committee members to update them with developments in the ethics of genomics research, handling of genomics information and biosafety requirements. Genetically modified food is one novel outcome of biotechnology with the potential to dramatically address the present food crisis in many African nations. It has become such a vexing issue with many political undertones that some African heads of governments have openly declared their countries' rejection of GM food aids.[27] [28] More often than not, most of these discussions are shaped and fueled by the opinions and writings of a few vocal and prominent individuals and/or organizations from inside but mostly from outside Africa. There is dire need for empirical qualitative and quantitative research to document people's perceptions and expectations about GM food and other potential future benefits of genomics revolution in Africa. Such research is required to provide basic knowledge about what the people actually want. It will also provide an opportunity to predict and proactively prepare for the social, cultural, religious and ethical implications of genomics to the African people.

Conclusion

The expected benefits from the human genome project and genomics technologies are fascinating and hold the ace for improving the standard of living of the African people. However, these expectations are futuristic, time-dependent and capital intensive. They require commitment of national governments to policy re-orientation about research and development, strategic planning, resource mobilization, priority setting, and establishing, promoting and sustaining enabling environments for scientific and technological breakthrough. No doubt, there is dire need for assistance from the developed and the frontline developing nations in form of information flow, developing educational capacity and technology transfer, but great initiatives and deep commitments must come from within. The time to begin is now as further delay would worsen the already existing gaps between the high and middle income nations on one hand and the low income nations on the other.

It should be acknowledged that biotechnology in its crude and traditional form is not new in Nigeria and other parts of Africa especially in some aspects of biology and agriculture. Moreover countries like South Africa and Egypt have advanced somewhat in modern biotechnology compared to others. However, there is the need to recognize and harness the revolutions and innovations that the knowledge of genomics has

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brought to molecular biology and biotechnology. Though genomics may not be the final solution to all the problems in which the developing countries are presently enmeshed, it might be a decisive and positive step towards solving most of their present and future health problems. In the words of Samuel Katz, "difficulties that present themselves now should not prevent us looking into future possibilities."[11]

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UK Biobank: a model for public engagement?

MAIRI LEVITT

Whilst in other applications of genetic technology the public debate has begun only when a piece of research has been completed, public consultations on biobanking began in 2000, before the funding for UK Biobank was even agreed, and have continued throughout its development. UK Biobank has obvious attractions for the British public. It is being set up specifically as a resource for research into common diseases that are relevant to everyone, rather than rare genetic disorders unknown to most. The only diseases mentioned on the 'about UK Biobank' web page are cancer, heart disease, diabetes and Alzheimer's disease[1]. The public are encouraged to be involved by the promise of 'a better life for our children and grandchildren' and 'enormous potential to result in improvements to health of the UK population' through the National Health Service.[2]

Ensuring public support

Despite these selling points it was recognised by the Medical Research Council (MRC) and Wellcome Trust from the start, that work would have to be done to ensure that UK Biobank would be a success[3]. The early consultations indicated reasons why public support could not be taken for granted. There was recognition of a problem of trust in science and science governance in the UK with the 'BSE crisis' and the media furore over GM food following the reporting of Pusztai's research with rats and GM potatoes and his concerns over GM food[4]. The first public consultation on UK Biobank stated that genetic research had 'a raft of unhelpful negative associations, based sometimes on misinformation and mistaken assumptions'[5] In contrast 'some people were better informed…and tended to have a more favourable view' [6]. UK Biobank has taken the view, found in many policy documents in the field of genetics, that transparency and openness is the key to increasing public confidence and trust [7]. The website contains a wealth of information including on-line minutes of meetings, results of public and stakeholder consultations, names and biographies of committee members and, with a push under the Freedom of Information Act, the reviewers' reports on the scientific protocol [8].

So UK Biobank might be seen as a model for public involvement having commissioned eight consultations with different groups between 2000 and 2003 and provided open access to the findings [9] . The public were consulted through surveys, focus groups and a people's panel before the plans were finalised; there were lay members on the committee devising the ethics and governance framework and on the committee devising the scientific protocol. Different publics were consulted throughout the process of planning the Biobank, for example, there was a consultation once the Ethics and Governance framework was devised and the views of those taking part in the pilot project are currently being gathered (the pilot began in October 2005).

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Tackling the ethics

Many of the traditional ethical concerns about medical research were avoided altogether by the protocol for UK Biobank. The age group chosen does not include children who cannot consent, would be unlikely[10] to include pregnant women or women planning a pregnancy, targets the currently healthy rather than vulnerable sick and avoids the elderly. The request to donate will be made independently of any treatment being received.

Other ethical boxes are ticked under the arrangements for the biobank. There will be:

- Voluntary participation
- Individual consent with general information on data uses and types of research
- Right to withdraw at any time
- No property rights over donated sample
- Access controlled using the existing system of research ethics committees
- An independent Ethics and Governance Council
- Security arrangements and assurances of confidentiality

What were the public *not* asked about?

The public were regularly consulted as the project developed in order to find out what would increase public interest and confidence and so ensure enough people would participate. Thus, in the early stages, consultations asked about general attitudes to genetic research and, later on, asked people to consider technical questions about how samples should be collected, issues of consent and access. The public were not invited to consider more fundamental questions about Biobank itself, for example, the priorities of commercial users versus the public interest , the likelihood of benefits set against other possible uses of those resources, the content of regulations and who would be enforcing them[11]. These sorts of concerns were covered by assurances that UK Biobank will 'Ensure that UK Biobank is used in the public interest', that as a charitable company 'it will only be allowed to act in the public good', that applications to use the samples would be subject to ethical scrutiny by research ethics committees and so on. As numerous studies have already shown, the public (or rather the diverse 'publics') have expertise from their lived experience that leads them to raise issues that may be overlooked by scientists, policy makers and others acting in their capacity of 'expert'[12]. For example, 'public benefit' may be a nice sounding phrase but who decides what is a public benefit, how are the public involved in the decisions and what happens when there are disputes about what is and is not a public benefit?

Power of veto

Power currently rests with the Board of Directors consisting of a healthcare policy expert, representatives of the funding bodies (Wellcome Trust, Medical Research Council and Department of Health), Professors of General Practice and Clinical Medicine and a chartered accountant. Not only are there no lay representatives, there are no ethicists or social scientists. The scientific committee has twelve professors in epidemiology, public health and other areas of medicine, an ex-nurse, the ex-chair of

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an NHS Trust and a professor of social policy. The Ethics and Governance Committee includes a marketing consultant, two people involved in consumer/patient interests, a barrister, a medical doctor, a professor of pharmacology and three ethical experts. This committee has been set up to safeguard donors and the public in general by reviewing the users of data and the types of research that are proposed. However, it is not the Ethics and Governance Committee that has the power of veto over the use of data or samples. This power belongs to the Board of Directors. A member of the scientific committee sits on the Board of Directors but there is no member from the Ethics and Governance Committee. This does not fulfil the promise in the Government White Paper (2003) that that there would be an 'independent monitoring body' with the power of veto i.e. presumably it was intended that such a body would be independent of the funding bodies [13].

If the Ethics and Governance Committee feels that a particular application is not in the public interest, or is unethical for any other reason, they can report publicly on their views. If they raise concerns and are not satisfied with the response from the Board of Directors they could resign. Given the acknowledgement in early UKBiobank consultations of the influence of the media in the field of genetics, this seems an unwise limitation. If the committee did 'go public' no doubt there would be extensive media coverage and a subsequent effect on recruitment/retention of donors.

Upstream but powerless to control the flow or dam the waters!

'Upstream' public engagement involves the public at all stages of scientific innovation and involves them in the direction of policy rather than simply inviting them to comment on existing arrangements [14]. In UK Biobank the public were involved 'upstream' in the sense of being involved early on in the project's progress but were not asked about the direction of the stream or its final destination. There was 'upstream' ethics engagement too, involving bioethicists, social scientists and other non-scientists whose task was to anticipate the boulders and other obstacles and smooth the flow of the stream. They are not able to control the direction of the river or to dam it if they feel it should be stopped. Both public and ethicists have played a part in smoothing the path of the stream but, under current plans, control lies elsewhere.

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[1] http://www.ukbiobank.ac.uk/about.php).

[2] Press release, 2002, UK Biobank website http://www.wellcome.ac.uk/doc_WTD002895.html

[3] Funding for the UK Biobank comes from the Medical Research Council; which is in funded by the UK Government; the Wellcome Trust which is a medical research charity, the Department of Health and the Scottish Executive.

[4] Horton R. (1999) Commentary: Genetically modified foods: 'absurd' concern or welcome dialogue? *Lancet* 354: 1315
Lancet (1999) Editorial:Health risks of genetically modified foods. *Lancet* 353:1811

[5] MRC/Wellcome Trust (2000) *Public perceptions of the collection of human biological samples.* Qualitative research to explore public perceptions of human biological samples Report prepared by Cragg Ross Dawson for the Wellcome Trust and Medical Research Council p.25f

[6] ibid

[7] Jones M. and Salter B. (2003) The governance of human genetics: policy discourse and constructions of public trust. *New Genetics and Society* 22:1 pp.21-41.

[8] See http://www.ukbiobank.ac.uk

[9] http://www.ukbiobank.ac.uk/ethics/consultations.php

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[10] Although the age range for participation has now been widened to 40-69 (from 45-69).

[11] All these sorts of concerns were raised in focus groups held with the general public (mixed age, sex, social background) discussing perceptions of privacy and trust in relation to personal medical and genetic data see Levitt M. and Weldon S. (2005) 'A well placed trust? Public perceptions of the governance of DNA databases' *Critical Public Health* 15:4

[12] For a summary of purpose, methods and activities see Sue Weldon (2004) *Public engagement in genetics: a review of current practice in the UK* http://www.cesagen.lancs.ac.uk/resources/papers.htm

[13] Department of Health (2003) *Our inheritance our future. Realising the potential of genetics in the NHS*
http://www.dh.gov.uk/PolicyAndGuidance/HealthAndSocialCareTopics/Genetics/GeneticsGeneralInformation/GeneticsGeneralArticle/fs/en?CONTENT_ID=4016430&chk=RnGBgL
The full paragraph reads 'An independent monitoring body will also be established to ensure that samples of genetic material are taken with the fully informed consent of the participants and that procedures to protect confidentiality are strictly adhered to. This body will have the power to veto uses of the data or samples that it considers to be against the interests of the participants or likely to damage the reputation of the study'.(para 5.37)

[14] Wilsden and Willis (2004) *See-through Science. Why public engagement needs to move upstream*. London: Demos

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Postgraduate Forum on Genetics and Society: Report on the Ninth Colloquium.

ANDREW BARTLETT, JAMIE LEWIS & INGRID HOLME

Initiated in 1998, the Postgraduate Forum on Genetics and Society (PFGS) provides a common forum for early-career academics from a variety of different fields, including sociology, psychology, law and philosophy, to engage with each other in productive dialogue. The annual PFGS Colloquium has a tradition of enabling those with interests in genetics and society to present their work to an audience of peers from a range of disciplines, traditions and institutions. These gatherings promote debate and dissemination in a rich inter-disciplinary setting and provide opportunities for cross-fertilisation of methods, theory and findings. They give the next generation of academics the opportunity to identify emerging research themes and the chance to attend free workshops, on such topics as methodology and ethics, led by established academics.

Cardiff University and CESAGen played host to the Ninth PFGS Colloquium between 31st August and 2nd September 2005, generously funded by the Genomics Forum. The title of the colloquium was 'The Genetic Information Age', an attempt to concisely articulate the central theme; a concern with the philosophical, scientific, legal and social implications of understanding genetic knowledge as information, or genetic information as knowledge. As with the PFGS, this theme was intended to be forward looking, urging consideration of the possible futures of the social and scientific landscape that current members of the PFGS will explore in the course of their careers. However, as Jon Turney argued when opening the colloquium with a keynote address, 'The Genetic Information Age' may well have already taken place.

Five themes were central to the postgraduate presentations:

1. The generation of genomic and post-genomic knowledge
2. The engagement of genomic science and scientists with the wider community
3. The development, structure and maintenance of intellectual property frameworks
4. The international context of genomics
5. The ideology of genetics

The generation of knowledge

A number of papers addressed the issues of scientific development, labour and organization. In the first session Andrew Bartlett (Cardiff University) presented a paper arguing that the development of 'big' biological science might have unintended effects on the generation of scientific knowledge. His paper suggests that a lack of attention has been paid to the hierarchical character of laboratory life within research. He argued that a scientific organisation maximising 'functional rationality' might lead to dissolution of the norms of the scientific community and an alienation of the scientists from the knowledge they create. In the same session, Miguel Garcia-Sancho (Imperial College) challenged the conventional account of the development of molecular sequencing strategies. He argued that the development of DNA sequencing

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was not simply a linear progression from protein sequencing that could be accounted for in purely technical terms. Garcia-Sancho argued that the development of DNA sequencing was directly influenced by the conceptualisation of DNA as information. This session gave us an image of knowledge production as an activity involving both the scientist as an individual, with personal norms and values, and the scientist as a member of a community, sharing concepts and metaphors.

The issue of how 'mutual' concepts and metaphors are created within scientific communities was explored by Bart Penders (Maastricht University). His paper stressed the different 'style boundaries' that exist between 'wet' scientists (biologists), and 'dry' scientists (bioinformaticians). Jamie Lewis (Cardiff University) further developed this issue, exploring the difference in the levels of standardisation in proteomics research, making reference to the Proteomics Standards Initiative, and between the dry and the wet laboratory, and the effects that this might have on knowledge production.

The role played by non-humans in the production of knowledge was emphasised in the paper presented by Shirlene Badger (University of Cambridge). This explored how, in the case of obesity, mouse models were used to directly inform research on children. These papers illustrate the methodological importance of qualitative studies of science with regard to understanding the process of knowledge production. This was articulated by Megan Clinch (London School of Economics) in a paper that called for a programme of 'narrating genes in context'.

Community engagement

'Community engagement' is here used as to describe the wider context of genomic knowledge, including public and political engagement with the rhetoric of genetics and genomics. Aryn Martin (Cornell University), who is in the closing stages of her PhD work, presented a paper that explored the concept of chimeras and mosaics. Her paper argued that cells have a materiality that can escape the body and enter institutions of governance, kinship and healthcare. Martin challenged the development of a one-to-one biographical correspondence between cells and people, presenting ethnographic case studies where the genetic variability of cells invokes a multiplication of personhood. Here, it is not so much that the science is new but rather that new people are constructed.

Connor Douglas (University of York) discussed the role of patients in co-constructing medical genetic technologies. He argued that the key issue here is not the ontological question of whether genetic information is of a different kind from other kinds of medical knowledge, but whether patients perceive genetic information as being categorically distinct. Ingrid Geesink (Cardiff University) presented a richly detailed paper that discussed the role of regulatory actors in shaping the technology of tissue engineering, with a particular emphasis on issues of risk and safety.

David Larsen (University of Cambridge) presented a paper that discussed the biopolitics of biotechnology. He argued that the primary interest of the British state in managing its citizen's illnesses is undergoing a process of realignment. Whereas the traditional model is for state intervention in the management of disease in order to

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increase the productivity of economic activities outside of the hospital and clinic, the emerging model is for the state to contribute to the endogenous growth of the biotechnology industry and their "reflexively" spiralling markets.

Intellectual property

It could be argued that the notion of genetic knowledge as intellectual property is a consequence of a perspective that holds genetics to be primarily an informational science. The key papers at the PFGS colloquium that addressed the theme of intellectual property were delivered in a themed session on the second day of colloquium. Chris Hamilton (London School of Economics) discussed the tension between the promissory, progressive rhetoric of 'bioprospecting' and the rhetoric of 'biopiracy' that stressed the inequalities of the relationships involved in 'bioprospecting'. This confrontation has found a site of particular contest in the arena of intellectual property rights; ought living things, and/or parts of living things, be patented?

Adam Bostanci (Exeter University) explored the arguments used by the U.S. Patent and Trademark Office (USPTO) to reject patent application on microbial genomes. He analysed the USPTO examination files relating to three applications to patent whole genomes. These were not deemed to be patentable inventions, whereas genes and open reading frames were. This paper raised questions of how divisions are drawn between the categories of invention and discovery, between an electronic genome sequence and its biochemical equivalent, and between a genome and a gene. Both these papers emphasised the changing boundaries between public and private agencies and raised issues surrounding responsibilities of institutional structures.

Several papers addressed issues of the general property structures that have developed with and are utilised by genetics and genomics. Both Larsen and Rebecca Hanlin (University of Edinburgh) discussed the importance of international property rights in modern healthcare systems. Adele Langlois (Open University) explored the impact of the inequalities in the proprietary structure of genetics and genomics in the global South.

International context

Langlois questioned whether existing initiatives, such as a series of UNESCO declarations and the Global Genomics Initiative, will be able to bridge what she describes as the 'genomics divide'. She asked whether the global South will be able to make use of the 'global public good' of genomics, or will it be the case that 'The Genetic Information Age' will produce greater inequality?

The international context of genetic knowledge and technology was explored by a number of other participants. Matthew Harsh (University of Edinburgh) discussed the governance of biotechnology in Kenya and Uganda. These nations have similar levels of biotechnology and similar biosafety policies. However, discourses of openness and mutuality present a more transparent and participatory image of biotechnological governance in Uganda than the governance regime employed in Kenya, which is marked by discourses of closed networks, specifically the 'Nairobi biotech mafia'.

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Chamu Kuppuswamy (University of Sheffield) addressed the 10/90 gap in terms of genomics. Following on from the Commission on Health Research for Development statement, in 1990, that only 10 per cent of the resources allocated for health research are directed to 90 per cent of the world's health problems, Kuppuswamy asked whether a similar pattern will be found in the distribution of the medical fruits of genomic science. She argued, illustrating her point with the case of AIDS drugs in South Africa, that inequities in healthcare are likely to be exacerbated by the advent of "The Information Age". Her colleague Yog Upadhyay (University of Sheffield) examined the North-South divide in agricultural biotechnology, arguing that the development of technologies shaped by the interests of the food industry of the North have a deleterious impact on agricultural practice and food security in the South.

The second day of the colloquium saw a session devoted to Canadian perspectives on 'The Genetic Information Age'. Anne Dijkstra (University of Twente) discussed the engagement of the wider society with genetic science, using divergent 'publics' as the object of her study rather than a homogenous 'public'. She compared debates on biotechnology and genomics in Canada with those taking place in the Netherlands. Grace Reid (Cardiff University) delivered a paper exploring social representation of cloning in Canada. A member of the Cardiff School of Journalism, Media and Cultural Studies, Reid presented the results of her textual analysis of newspaper articles on cloning and a series of focus groups. She discussed the framing of the benefits of cloning and how these are weighed against moral and ethical concerns.

The ideology of genetics

The final session of postgraduate presentations discussed the idea of an 'ideology of genetics'. Kean Birch (University of Glasgow/Oxford Brookes University) discussed the development of ideological networks within and around the life science industries wedded to 'economic' rather than 'technical' success. Jane Miller (University of Sheffield) asked if the future of medicine was genetic. She pointed out that historically, major improvements in health care have been the result of social programmes, and that genetics is just a small part of our health care requirements. Miller suggested that political enthrallment with the promises of genetic knowledge could have serious consequences if this entails a diversion from conventional public health strategies. Kate Weiner (University of Nottingham) delivered a critique of the notion of geneticization, a concept that has been used across a wide range of research since the early 1990s. Her paper used the work of PFGS alumni Adam Hedgecoe on the rhetoric of science to suggest that we need to consider how we perform and disseminate critical research on genetics while not playing an active role in the construction of 'hype'. Birch drolly proposed that we ought to encourage others to ignore our work so as not to contribute to the 'hype'.

Conclusion

The Ninth PFGS Colloquium attracted 51 attendees over the three days, including the guest speakers; Jon Turney (University College London) who opened the colloquium, Andrew Webster (University of York) who led a research ethics workshop, Kate Stewart and Bruce Mason (both Cardiff University) who led a pair of themed training workshops on the use of internet communication in social research, and Joan Haran (Cardiff University) who provided an after-dinner speech on the evening of the

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second day. The colloquium closed with a panel discussion on the notion of 'The Genetic Information Age' involving Ian Brewis (Cardiff University), Jane Calvert (Exeter University), Katie Featherstone (Cardiff University) and Buddug Williams (Genetic Interest Group). The key measures of the colloquium's success however are the wealth and quality of the postgraduate papers that were presented.

30 postgraduate papers were delivered. While the papers discussed in this review are an attempt to provide a representative picture, the full set of colloquium abstracts can be found on the PFGS website.

For further information

The PFGS manages a website at http://pfgs.org/ and a bulletin board at http://pfgs.org/phpBB/index.php. The website contains information on previous colloquia, member profiles and details of upcoming events. The bulletin board is a site for direct discussion of issues of interest to researchers working in the field of genetics and society. To become a member of the PFGS please visit the website or contact any of the authors of this paper. Membership is free.

Note from the Genomics, Society and Policy Editors:

There will be a PFGS Special Issue of the Genomics, Society and Policy Journal published in December 2006, which will feature papers related to the theme of the Ninth PFGS Colloquium, The Genetic Information Age.

Any enquiries can be directed to PFGS Co-Editor, Adam Bostanci, at a.w.s.bostanci@ex.ac.uk.

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David Winickoff is Assistant Professor of Bioethics and Society at University of California , Berkeley. He has also taught at the J.F.K. School of Government at Harvard University and he holds degrees from Yale University, Cambridge University (U.K.), and Harvard Law School. His work focuses on the ethics, law and governance of biotechnology.

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