Debate

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DNA databanks and consent: A suggested policy option involving an authorization model

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Abstract

Background

Genetic databases are becoming increasingly common as a means of determining the relationship between lifestyle, environmental exposures and genetic diseases. These databases rely on large numbers of research subjects contributing their genetic material to successfully explore the genetic basis of disease. However, as all possible research questions that can be posed of the data are unknown, an unresolved ethical issue is the status of informed consent for future research uses of genetic material.

Discussion

In this paper, we discuss the difficulties of an informed consent model for future ineffable uses of genetic data. We argue that variations on consent, such as presumed consent, blanket consent or constructed consent fail to meet the standards required by current informed consent doctrine and are distortions of the original concept. In this paper, we propose the concept of an authorization model whereby participants in genetic data banks are able to exercise a certain amount of control over future uses of genetic data. We argue this preserves the autonomy of individuals at the same

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time as allowing them to give permission and discretion to researchers for certain types of research.

Summary

The authorization model represents a step forward in the debate about informed consent in genetic databases. The move towards an authorization model would require changes in the regulatory and legislative environments. Additionally, empirical support of the utility and acceptability of authorization is required.

Background

Recent developments in genetics, particularly the sequencing of the human genome, have energized large-scale genetics and genomics research. One of the outcomes has been the establishment of large-scale genetic data banks aiming to identify genetic predispositions to major public health conditions that appear to have complex associations rather than being caused by single genetic mutations. Although many small collections have existed for a long time, none have been on a massive national scale until recently, when a private Icelandic company working closely with the government of Iceland established the Icelandic genetics database [1]. Specific legislation had to be passed to enable the creation of that database. Since then various countries, including Estonia, have attempted to establish their own national data banks. The United Kingdom is now in the process of creating the world's largest such bank. It will be known as Biobank and will collect DNA samples from approximately half a million adults [2].

An important feature of these data banks, and one that is crucial to their scientific utility, is the ability to link DNA information with individual clinical outcome data and, perhaps, relevant non-medical information. As such, these data banks create profound legal and ethical issues, especially in areas of privacy, confidentiality and access [3-5]. In the conventional research setting, these issues are usually dealt with by obtaining informed consent for the use of individual health information for research purposes. But while it is possible to obtain informed consent to have one's blood, cells or tissue sample taken by the researchers for a specific research project, the very intention of setting up such large data banks precludes giving informed consent for all the possible ways in which the information derived from that sample can be used for future research [6]. Given the speed of scientific development in the area of genetics and the vast spectrum of potential research hypotheses that may arise and can legitimately be addressed by such databanks, there is no way to predict possible future uses of donated samples.

In this brief article, we do not propose to resolve the policy dilemma. Rather, we mean to highlight that much of the existing literature and policy statements underplay the stark nature of the relevant policy choices. In the context of research involving large-scale DNA databanks, it will be nearly impossible to craft a policy solution that can meaningfully satisfy existing consent norms. The choice for the public and policy makers is between the research and an abandonment of existing consent principles. We will discuss an authorization model that may help to clarify some of the tensions inherent in consent models.

Discussion

Informed consent

The modern understanding of informed consent in the context of research is greatly informed by the Nuremburg code and the large body of ethics literature and analysis that has emerged since World War II [7]. At the current time, the legal obligation of informed consent in the research setting is tremendously onerous and has been characterized as "the most exacting duty possible" [8]. At a minimum, it requires researchers to provide information about all potential risks, no matter how remote, and material information about the nature of the research protocol. In the context of clinical genetic research, this consent process should include, for instance, information about possible commercialization, how issues of confidentiality will be addressed, potential impact of participation on insurability, and whether the research results will be available to the research

participant. In the context of clinical research, few would dispute the value and necessity of this robust consent process. However, should the same rules apply to research involving DNA data banks?

Though there are some jurisdictions that have legal frameworks capable of allowing access to identifiable health information without consent (e.g., the UK's Health and Social Care Act 2001), as a general rule the law compels researchers to obtain informed consent for the use of identifiable health information, including tissue samples. This is because the law views health information as something that, as suggested by the Supreme Court of Canada, "goes to the personal integrity and autonomy of the patient" [9]. And, as such, the patient maintains a "basic and continuing interest in what happens to this information, and in controlling access to it." [9].

But if we are to allow population genetic research to move forward, we need to recognize that adherence to this traditional model of informed consent is problematic. As the UK Human Genetics Commission recently concluded: "the difficulties involved in tracing and securing re-consent for different forms of medical research may make obtaining fresh consent impractical and would seriously limit the usefulness of large-scale population databases" [10].

Population research does have characteristics that seem to justify a re-assessment of the consent standards. First, as noted, the purpose and direction of the research may not be fully known at the time the samples and consent are obtained. Second, because much of the research involves low penetrance genes, it is unlikely the research results will be of immediate clinical relevance to individual research participants [11]. Third, the research and information involves large numbers of people, thereby minimizing the impact and relevance of a single sample. Fourth, the DNA samples can be collected relatively easily and involves little physical risk. And finally, the research may require multiple requests for consent, thus burdening both researcher and participant, which was noted on a recent public consultation to be a potential disincentive to participation [12].

Nevertheless, in total, it is difficult to argue that this is research that does not require, at least technically, specific consent [13]. Consent law is concerned with providing research participants with relevant information in order to allow autonomous decision-making. Withholding or tailoring the provision of information in order to meet a broader social agenda, conflicts directly with the ethical principles that underlie much consent jurisprudence. Moreover, there are aspects of population genetic research that seem especially important to communicate as part of an ongoing consent process. For example, the collection and storage of the DNA samples, particularly when it involves discrete or identifiable populations, has the potential to create a variety of social and ethical concerns, including possible genetic discrimination [14]. In addition, it might have health and legal implications to third parties, particularly genetic relatives across several generations. And, rightly or not, research participants may also have a continuing interest in the commercial applications associated with the DNA databank [15].

Policy options

The issue of consent in the context of DNA data banks has been the subject of a variety of recent policy documents [10]. Though there seems to be an emerging consensus regarding the nature of the consent challenges created by DNA data banks, most policy recommendations continue to rely on an unworkable straining of existing consent principles [16]. The most common recommendation seems to be the adoption of some form of "blanket consent", that is, a consent form that allows research participants to make a one-time choice about the future use of their DNA sample. For example, UNESCO's *Draft Report on Collection, Treatment, Storage and Use of Genetic Data* recommends: "A system which required fresh consent would be extremely cumbersome and could seriously inhibit research .... blanket consent covering all forms of future medical research might be preferable [17]. Similarly, commentators have also suggested using a system of "presumed consent." Based on the results of a study which found only 29% of the survey participants would want to re-consent, Wendler and Emanuel suggest "using presumed consent with opt-out" [18].

Though a one-time consent would undoubtedly simplify the research process, blanket consents cannot be considered true consent. Because blanket consents are necessarily vague, they are, by

definition, far too general to have much legal weight. Moreover, they do not allow patients to meaningfully act on their continuing interest in their health information [19]. As such, most types of blanket consent will, as suggested by Hank Greely, fall "far short of true informed consent" [20]. A variety of studies of public opinion have demonstrated a strong desire for a retention of the consent process in this context [21-23]. For example, a study done by the UK Human Genetics Commission found that 82% of the respondents either strongly agree (44%) or tend to agree (38%) that fresh consent must be sought from individuals before new research can be conducted on existing DNA samples held in medical genetic databases [24].

An alternative is to move away from the "fiction of consent" [13]and recognize that a new legal/ethical framework is required. For example, a more appropriate model may be that of authorization. Greely has set out a series of issues that must be addressed in order valid future research on collected genetic samples. His model includes permission for unforeseen research, recontact of subjects, the right to withdrawal, setting time limits on the use of samples, availability of information or materials to third parties, information on implications for groups and information on commercial uses [20].

Whereas individual informed consent would be required for initial collection of genetic material and health information, subsequent uses could be carried out under a mode of pre-authorization. This could take the form of a directive, such as a proposed health information directive that gives participating individuals the ability to pre-specify uses for which they do or do not wish to give informed consent in the future [25]. For genetic data banks, participants may wish only to be contacted if there are clinically relevant findings, or if potential commercial applications are being derived. The possibility of a blanket consent exists, but is not presumed by researchers and can only occur by the choice of a participant. Each individual can specify in advance the extent of involvement with decision making that is desired. This preserves aspects of autonomy, but is neither restrictive of future uses as a full consent model, nor is as permissive as proposed blanket consent models. Recent research indicates that participants are willing to consent to research when contacted many years after the original collection of genetic material, and most would do so if an ethics review board had approved such studies [26]. Despite this, it would still be preferable to have such a process set out in advance rather than always working retrospectively.

Such an authorization model would need to be structured with an understanding of the protections and fundamental rights that are lost through a change of the consent process – including an understanding of the social and ethical concerns specific to this area of research (e.g., concern about discrimination and stigmatization). For example, the system would need to be bolstered by additional protection afforded by an overarching governance framework of trust, responsibility and accountability. The involvement of institutional review boards would be essential. Additionally, a socially constituted, preferably legally mandated, oversight body operating at arms length from researchers and commercial interests such as the Medical Data Panel proposed by the Select Committee on Science and Technology in the UK, or an ombudsperson, such as that proposed for electronic health information, would bolster these additional protections. In many jurisdictions, such as in Canada, the adoption of an authorization model or, for that matter, any scheme that differs from the existing consent principles, would require the enactment or amendment of legislation.

Summary

The value of recognizing that existing consent norms are incapable of accommodating much of the research associated with DNA data banks is that it forces policy makers and the public to confront the social tradeoffs inextricably linked to this work. If we are to adhere to the well-established consent norms, a good deal of population research may not occur. On the other hand, if we abandon the current consent model, research participants will be giving up well established rights and a degree of control. By recognizing the choice, society can more clearly debate the benefits and risks of each course of action.

Competing interests

None declared.

Authors' contributions

AD, REGU jointly initiated the idea paper. AD, REGU and TC contributed to the drafting and subsequent revision of the paper. All authors read and approved the final manuscript.

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