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GOVERNING GENETIC DATABASES: COLLECTION, STORAGE AND USE

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Abstract

This paper provides an introduction to a collection of five papers, published as a special symposium journal issue, under the title: “Governing Genetic Databases: Collection, Storage and Use”. It begins by setting the scene, to provide a backdrop and context for the papers. It describes the evolving scientific landscape around genetic databases and genomic research, particularly within the biomedical and criminal forensic investigation fields. It notes the lack of any clear, coherent or coordinated legal governance regime, either at the national or international level. It then identifies and reflects on key cross-cutting issues and themes that emerge from the five papers, in particular: terminology and definitions; consent; special concerns around population genetic databases (biobanks) and forensic databases; international harmonisation; data protection; data access; boundary-setting; governance; and issues around balancing individual interests against public good values.

A. INTRODUCTION—SETTING THE SCENE

Over the past decade, human genetics research has undergone extensive and rapid growth, both in the UK and worldwide. The avenues of inquiry now open have expanded dramatically, as major advances in high-throughput DNA sequencing, genomic research methods, information technology, computing and associated biotechnologies have increased substantially the range of tools and techniques available to scientists. One of the most significant developments has been a burgeoning in the number and sophistication of collections of human genetic materials used for biomedical research or other purposes, including criminal forensic investigations.1 Such collections—which, for convenience, we shall refer to as “genetic databases”—cover a kaleidoscopic range of forms. However, in essence, they tend to be systematically organised collections that typically store human tissue, bodily samples, extracted DNA and/or other physical genetic material, together with relevant personal, medical, genealogical, genetic and/or lifestyle data, on increasingly large numbers of individual participants.

Traditionally, most genetic databases have been relatively small-scale collections of biosamples and data, held by individual clinicians, research groups or institutions, and established to elucidate the genes and genetic variations involved in single diseases. Recently, though, the global genetic database landscape has changed fundamentally.2 Within the biomedical realm, several countries have taken steps to create population-scale genetic databases. These highly ambitious initiatives—of which UK Biobank is a leading example3—seek to establish long-term, multi-purpose repositories of genetic biosamples and data, obtained from many hundreds of thousands (if not millions) of individuals. Rather than being limited to single diseases or specific, short-term projects, these so-called

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"population genetic databases" or "biobanks" are designed to support a huge range of genetic, biochemical and epidemiological research into common disorders that involve complex gene-environment-lifestyle interactions.4 Over the course of several decades, population genetic databases are intended to be accessed and used by any number of different researchers, potentially located anywhere around the globe, whether based in the public, charitable or commercial sectors, and who may be engaged in as yet unknown and entirely unforeseeable kinds of research.

Alongside the emergence of population biobanking, moves also are gathering pace to network, interlink or integrate existing genetic databases, both within countries and on a multinational scale. There is the potential for multiple, distributed datasets to be integrated, searched or shared as if they were single, unified collections, in order to harness and exploit the benefits of vastly increased statistical power. This is particularly vital for genomic and epidemiological research, including genome-wide association studies.5 Various consortia with members based in the UK—notably, the P^3G Consortium6—have begun to emerge, having the aim of facilitating such networking and promoting international collaboration.7 Their mission includes identifying core legal and ethical norms and principles that should guide and inform genetic research policies and practices, as well as developing harmonised standards and procedures, particularly for transborder data-sharing and securing research ethics approval.

In the forensic science realm, too, parallel initiatives can be seen. While the UK ranks second only to the USA in terms of biomedical research excellence,8 in criminal forensic intelligence database terms it leads the world. The National DNA Database®, established by the police in England and Wales, is easily the largest genetic database of its kind in the world. During the 11 years since its inception, it has amassed nearly 4 million DNA profiles on individuals.9 Meanwhile, as in the biomedical sphere, UK police forces are actively exploring the possibility of linking up their intelligence and forensic collections with other countries' equivalent databases. This provides enormous possibilities to track individuals across jurisdictions and to address global criminal activity. But the establishment and growth of large genetic databases established for medical research purposes also increasingly could become attractive to the police and other security agencies, as a means of identifying suspects or obtaining comparative DNA samples. Currently, it is very difficult to deny police access to any biomedical collection; access can be achieved by application for a court order in most jurisdictions.

All of these developments—the burgeoning of genetic and genomic research, proliferation of genetic databases of all shapes and sizes, rapid technological strides, and mounting pressure for international collaboration and data-sharing—bring into sharp relief the need for an appropriate and effective framework for governing genetic databases, both at the national and international levels. This is crucial, not only to safeguard important rights, interests, principles and values, but to facilitate beneficial research for the public good and to maintain public confidence and support. Yet, as Bartha Maria Knoppers *et al.* demonstrate in their contribution to this special journal issue which maps the existing national and international governance frameworks and supervisory bodies (such as they are), current governance and oversight provisions fall well short of the mark. As long ago as 1994, the House of Commons Science and Technology Committee described human genetics as a field "ripe for review and regulation".10 Nevertheless, to date we still remain without any clear, coordinated or coherent framework for governing genetic databases and related activities, even at the domestic level. Perhaps unsurprisingly, then, issues central to genetic database governance have attracted growing attention from commentators. Increasingly, policy-makers and lawmakers, too, are recognising the urgent need for sound and robust governance provision. Not least, this is because—as all of the essays in this special issue

amply demonstrate—genetic databases raise a host of challenging issues, many of which test our traditional legal concepts, governance provisions and bioethical principles.

B. CROSS-CUTTING CHALLENGES AND THEMES

As all five papers in this special collection show, genetic databases pose numerous challenges and questions. Many of these are novel and somewhat unique to human genetic databases. One of the fundamental issues that remains unresolved is reaching a common *definition* and agreement on *terminology*. While, for convenience, we have favoured "genetic databases", such collections are in fact known by a veritable smorgasbord of different terms. Other common labels include biobanks, tissue banks, human genetics research databases, genomic databases, population databases, research repositories, DNA databanks and gene banks—as well as variations along the same themes.11 To some extent, as Tim Caulfield notes in his paper, such variations in terminology mirror the heterogeneous nature of collections. For, the "genetic database" universe encompasses a vast spectrum, including collections that may differ significantly in their size, origins, purposes for creation and/or use, design features, degree of technical sophistication, content, duration, range of users and ownership. But, as the paper by Knoppers *et al.* suggests, without some broad consensus over terminology (or, at the very least, standardisation around a limited range of accepted terms), and without achieving deeper conceptual clarity over how different collections should be defined and classified according to relevant type, framing effective, proportionate and appropriate governance mechanisms will remain an impossible dream. Both at the national and international levels, the law—as well as regulatees under it—must be able to say not only which entities, activities and actors are subject to regulation (and which are not), but when, how, and to what extent.

The issues that are raised by human genetic databases also challenge many of the fundamental principles and assumptions upon which traditional standards and procedures for medical research have been built. Many of these issues are novel and somewhat unique to human genetic databases. How those challenges and questions are answered may have significant implications, not only for stakeholders, policy-makers and lawmakers, but also—as Deryck Beyleveld and Roger Brownsword especially note in their essays—for the very nature of democratic society and moral community. In his paper, Caulfield helpfully captures a sense of the scale of the issues at stake by providing an indicative list. His list includes concerns around ownership; governance; community participation; providing feedback to participants; data-sharing; and third party access. Further key issues that are highlighted and discussed by the other contributors include: IT security; privacy, confidentiality and data protection; controlling data access and access to biosamples; benefit sharing; commercialisation; managing intellectual property rights; genetic exceptionalism; genetic discrimination; boundary-setting (in terms of novel database uses and database interlinking); balancing potential conflicts between different values or interests (especially the public good vs individual interests); and international consensus-building and harmonisation.

However, as Caulfield and Campbell each point out, it is issues around obtaining *consent* from genetic database participants that have attracted by far the greatest attention to date. Indeed, all five papers presented here address or touch upon various aspects of consent—from its ethical foundations, particularly in autonomy, and the implications of this (Alastair Campbell and Caulfield); through the centrality of consent under data protection law, the relevance of anonymisation, and the justifiability of data protection exemptions from consent for medical and genetics research in the public interest (Beyleveld); to possible arguments in favour of compulsory, universal participation in national genetic databases that serve both forensic and public health purposes (Brownsword). As all five papers show,

consent is particularly problematic in the *population* genetic database (or "biobanking") context. This is because the traditional prerequisites for valid, specific, informed consent from medical research participants to take and use their tissue, biosamples and data—especially for future, as yet unknown secondary research purposes—cannot be met. Within the ongoing international debate over the legitimacy (or otherwise) of "blanket" or "generic" forms of consent, it is illuminating to compare and contrast the differing perspectives put forward by Knoppers *et al.*, Caulfield and Campbell.

Consent—especially its proper form, and when and where it should be required—is a perennial issue within the wider literature that is dealt with in different ways by the authors in this series. As mentioned earlier, Brownsword regards the facility for exercising individual (moral) choice as being essential to the very fabric of a moral community. He is therefore against compulsory inclusion of individuals in genetic databases used for public health purposes, and regards consent as a prerequisite for their inclusion. Beyleveld gives us a better idea of when and where consent should be required in the context of genetic databases. He argues that explicit consent is required, even for the anonymisation of data, unless the processing of data (as provided for by law) would threaten more important rights of others, or be necessary for the protection of health in a proportionate way. Campbell, on the other hand, is much happier with a "broad" consent regime for inclusion in population biobanks. In his view, requiring that individuals must reconsent to specific subsequent uses of their personal information or biosamples would be "paternalistic". Instead, Campbell believes, the key criterion is trust: if a biobanking institution is trusted (and trustworthy), then it should be able to carry out any subsequent research within the scope of the original "broad" consent, subject to appropriate controls, without any need either to consult with, or ask the permission of, individual participants. Caulfield is reluctant to support such a view. He argues that consent should not be jettisoned simply because it is thought too costly or too difficult to obtain. Instead, he warns against succumbing to arguments favouring broad consent that are not based upon (or, at least, not reconciled with) foundational principles. In his view, the requirement of consent for medical research is grounded in fundamental human rights and rests on autonomy. Therefore, any alteration, amendment, weakening or abandonment of the traditional informed consent principle should be done only with careful understanding of the implications. So then, while all authors agree that there should be consent, their papers reflect a considerable and fascinating spectrum of opinion over its proper form and when it should be required.

As we find ourselves on the cusp of a new generation of large-scale, international genomic research collaborations, the question of global—or, at least, regional—*harmonisation* is becoming particularly pressing. Knoppers *et al.* focus on several key issues around harmonisation, including roadblocks to international collaborative research and potential means for overcoming them. They single out for closer scrutiny two specific concerns around which at least some internationally accepted norms have emerged—namely, benefit sharing and controlling access to data by third parties, especially external researchers. Beyleveld, too, focuses on issues around *data protection* and *data access*. In his case, it is to illuminate another thorny (and legally complex) area—namely, that of trying to strike a fair and proportionate balance between individual privacy values and medical research values under data protection laws, especially where the two might appear, at first glance at least, to pull in opposing directions. Drawing together both legal and philosophical source materials, Beyleveld's paper offers a fresh and penetrating analysis of the interface between data protection/privacy values on the one hand, and public interest/medical research values on the other. His conclusion—that their relationship may better be viewed as being "co-operative" rather than necessarily "conflictual"—may prove to be just as surprising to some readers as are the implications of the logical thought process through which Brownsword guides us in his paper.

Also drawing upon legal and philosophical tools, Brownsword's paper tackles head on important emerging issues about how, where and why we should set *boundaries or limits* around genetic database uses, both within the public health and the criminal intelligence and forensic spheres. In short, he asks: can compulsory, universal participation in a single national (or, conceivably, international) genetic database, that government(s) can use for (potentially any) "public interest" purposes, be justified? As genetic databases proliferate and our technological capacity to network them accelerates, sooner or later policy-makers and lawmakers will have to grapple with these and related questions. As Brownsword argues —and Beyleveld and Campbell, too, make similar points in their essays—what we decide may well have serious ramifications, not only for individual stakeholders and the overall public good, but for the very nature of the society within which we live.

While the principal focus of this special collection of papers is on human genetic databases used for biomedical research purposes, both Brownsword and Campbell examine *forensic* databases in considerable depth. At a time when the use of bioinformation for forensic purposes, and the governance thereof, are actively being reviewed in the UK,12 these discussions are timely and apposite. Significantly, both contributors (and Caulfield, too) caution against the danger that supposed "public good" justifications potentially could become a licence to commandeer or exploit genetic databases in ways that improperly sacrifice individual rights and interests. These papers demonstrate that the conceptual basis of the notion of the "public good" requires further development and analysis, particularly in the area of biobanking. It is evident that further debate and scholarship is required as to when the "public good" concept properly can be invoked and used as a justification for deviating from accepted principles of medical research and human rights law. The authors' calls for full public debate, and to embed within the governance framework now all necessary safeguards, oversight mechanisms and accountability structures to prevent future misuses or abuse, arguably should be heeded as a matter of urgency. Having said this, again, it is instructive to bear in mind Beyleveld's observation that apparent tensions between individual and corporate rights, interests and values need not always, in fact, be as "conflictual" as they appear, but that respect for human rights enhances medical research.

C. CONCLUDING REMARKS AND ACKNOWLEDGEMENTS

Overall, the five papers collected here stand as testament not only to the importance of fostering and facilitating beneficial research for the public good using genetic databases to the full, but also to the wide range of challenging issues and questions that any properly constructed genetic database governance framework ought to address. Taken together, the five complementary essays go a considerable way in advancing our awareness and understanding of many key issues—and, perhaps even more significantly, towards identifying possible ways to progress both the domestic and global quests to formulate practically workable, normatively sound, appropriate and effective governance mechanisms.

The five papers presented here were originally given as seminar presentations, as part of a seminar series on governing genetic databases held at the Ethox Centre, University of Oxford, during 2006. As co-convenors of that seminar series, and co-editors of the articles in this special symposium journal issue collection, we are indebted to all of the participants for giving so generously of their time and energy. Grateful thanks also to the Ethox Centre and the Oxford Genetics Knowledge Park for hosting and supporting the seminar series, and to Michael Parker and all of our colleagues at the Ethox Centre. The idea for this series emerged from research conducted in the Governing Genetic Databases project (2006-2008) that is funded under the Wellcome Trust's Biomedical Ethics programme (award ref: 076070/Z/04/Z). Finally, we wish to thank the present and former editors of the *King's*

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