

## **HUMAN GENETIC DATABASES: ETHICAL, LEGAL AND SOCIAL ISSUES**

### **Preface to the special issue of TRAMES**

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#### **1. The boom of DNA banking**

The last few years have witnessed an important expansion of DNA banking all over the world. The collections of DNA samples vary in design and purpose (Cambon-Thomsen et al; 2003 Palmour 2003), occurring in a variety of circumstances from research to diagnostic and therapeutic activities, as well as in forensic services in identifying individuals through DNA. Most of these DNA banks are of small size, mostly set up in order to enable research in the context of disease studies. The very recent tendency to build up extensive population-based databases is related to the success of the Human Genome Project that has energized large-scale genetics and genomics research.

While the mapping of the human genome has been a major scientific achievement, there still exists a large gap between gene discovery and our ability to utilize genetic information to improve health and prevent disease. The interest in human genetic variation and genetic epidemiology provide the basis for the construction of genetic databases. One hopes that the large-scale genetic databases enable us to understand the combined effects of genetic, lifestyle and environmental risk factors in the development of a disease.

Also emerging areas of research, such as pharmacogenetics, also require access to large pools of genetic data. Pharmacogenetics, as the study of genetic variation that affects response to medical drugs, has the potential to improve the safety and efficacy of treatments, mostly by supporting the development of genetic tests that would allow to judge how likely a specific medicine is to help or harm a particular patient. People vary in their response to the same medicine due to the differences in their genetic make-up. The hope is that pharmacogenetics will enable to find the right medicine to the right patient in the right dosis.

## 2. What is a population-based genetic database?

The population-based genetic databases rely on a large number of research subjects contributing their DNA samples in the form of blood or tissue that will be linked with medical, genealogical and lifestyle information.

Iceland has been a pioneer in proposing a database complex consisting of Health Sector Database, genealogical database (“Book of Icelanders”) and Genetic Database. This was followed by the Estonian Genome Project (Estonia), UK BioBank (United Kingdom), CARTaGENE (Quebec, Canada), UmanGenomics (Västerbotten, Sweden), Genome Database of the Latvian Population (Latvia), Genome Institute of Singapore (Singapore), Autogen Limited (Kingdom of Tonga). Also in USA and China projects are currently under way. The best overview of the planned genetic database projects can be found in Austin, Harding, McElroy 2003 and Cardinal, Deschênes 2003. It seems that all currently proposed population-based genetic database projects have the same goal – they intend to identify susceptibility genes for common diseases and attempt to improve the medical care and health of the populations involved. In some cases, like Iceland, Estonia and Latvia, the initiators of the projects also hope to boost the country’s economy through expanding the biotechnology sector and creating new jobs. Although the database projects share the main objectives, they vary in size, subject participation, organization, as well as in the balance of government and commercial involvement. The planned projects have different consent procedures and only some (Estonia, Latvia) intend to give feedback to the participants.

Researchers, physicians, patients, biotechnology firms and pharmaceutical companies are excited about the scientific and therapeutic potential presented by genetic databases. They are all interested in the discovery of the genetic causes of diseases and in the development of better treatments and cures. But their interests and motives for participating in genetic research are, however, different or even competing. The key interests characterizing the contributions of firms, foundations, governments, researchers, patient groups and universities have been very well outlined by Merz, Magnus, Cho, and Caplan (2002: 967). The researchers may be motivated by intellectual curiosity or self-esteem, the biotechnology firms and pharmaceutical companies are interested in financial return, the patients and their families can be motivated by self-interest in treatment or cure for a disease, but also by altruism or even duty.

Different interests are entangled here which means that the risks and benefits arising from the project should be analysed on very different levels. Besides potential benefits and harms to the individuals one should also take into consideration the implications for groups and communities involved. Besides liberal individual rights-based ethics there is an increasing interest in the communitarian ethics involving participation in research for the common good and the sharing of the benefits of research based on solidarity (Chadwick and Berg 2001).

Traditionally bioethics has operated with the notions of individuals whereas databases deal with large collectives. The different situations might require

different types of concepts and rules. The most important ethical challenge of national genetic databases is the need to reconcile the discourse of individual rights (privacy, confidentiality, right to know/not to know) with the discourse of collective rights and solidarity (genetic database as a common public good, duty to participate in research, duty to know and inform others).

The ethical challenges that are associated with genetic databases have attracted much attention from world organizations such as United Nations Education, Scientific and Cultural Organization (UNESCO), Human Genome Organization (HUGO), World Health Organization (WHO), and World Medical Association (WMA) (see “Documents” at the end) but no uniform general ethical and legal guidelines exist for building up and operating the population-based genetic databases.

### **3. Ethical, legal and social aspects of human genetic databases**

An important feature of genetic databanks is their ability to link DNA information with individual medical information and genealogical data. As such, these databanks create profound ethical and legal issues, especially in areas of ownership, privacy, consent, confidentiality and access. Several of these issues have been raised already in the relation to genetic testing, screening and research but have acquired novel aspects in the context of large-scale databases. What is new, is that it is not solely the individual but also the community that needs protection and should be asked for consent.

Several authors have suggested that the established concept of informed consent based on the international documents and regulations (Nuremberg Code and Helsinki Declaration) may not be applicable in the context of genetic databases where the future potential uses of genetic materials are not known at the time the consent is given. There is a discussion whether blanket consent, suggested by the World Health Organization in 1997, suffices (Deschênes, et al 2001, O’Neill 2001, Árnason 2004). Different proposals such as an open consent, a two-stage consent (public and individual consent), written authorization have been made.

Consent of the community is by far the most central ethical concern of large genetic databases. However, in several countries one hears complains about the lack of public debate. In some societies, like the UK, the database projects have been widely discussed by different interest groups (for example Genewatch UK) whereas in other societies only single individuals have expressed their reservations about the project (as it has happened in Estonia). The Tonga database project was terminated because of great opposition from church and pro-democracy groups. An Australian genomics company Autogen Limited planned to establish a database of genetic information of the population of 100 000 people of the Kingdom of Tonga. Although public consultation is not a legal requirement, it is understood to be an essential part of the informed consent process. How public consultation and public debate can be initiated is still unclear.

The major role in the public debate is played by the media. Whose voice is heard in the media? What kinds of arguments prevail in the media discourse on human genetic databases? Several articles in this issue of “Trames” show that the fact of existing media coverage does not always result in people actually understanding what is going on. The fact that in spite of the extensive media coverage in both Iceland and Estonia, only 7% of the Estonian population and 14% of the Icelandic population consider themselves to be well-informed about the gene bank project, makes one wonder whether it is at all possible to inform the general public about all aspects of such many-faceted large projects. How should science communication and science policy be organized?

Some of the most critical social issues raised by the databases are the questions of social justice and benefit sharing. The HUGO Ethics Committee suggests in its Statement on Human Genomic Databases (2003) that databases should be regarded as global public goods based on the recognition of the human genome as belonging to the humanity. The object of discussion is who and for what reasons has the right to require benefits arising from research and innovations. The rapid developments in the field of biomedical science and biotechnology promise huge advantages in medical treatment and healthcare. But from the perspective of social justice concerns have been raised as regards to the access to these sources and goods: the affluent populations could reap the rewards while the poorer countries will not be able to benefit due to the high price of new drugs and therapies. In order to protect the social welfare of these populations which have become the source of commercial genetic information, the HUGO Ethics Committee has suggested in its Statement on Benefit Sharing (2000) that a percentage (1%-3%) of the annual net profits of the entities responsible for genetic research should go back to these communities (for example through investments into the healthcare infrastructure).

#### 4. The general description of the special issue of “Trames”

The current issue of “Trames” casts some light onto the discussions concerning these questions. It is divided into two parts: the first part examines the ethical, legal, and social justice issues of databanks – ownership, informed consent, benefit sharing, feedback and access to the data. The second part deals with the social issues: public attitudes and debate, media discourse, science communication and policy.

The first article acquaints the reader with the preliminary results of the lawyers’ group work, which has been carried out as a part of the project “The Ethical, Legal and Social Aspects of Human Genetic Databases. A European Comparison” (ELSAGEN) financed between 2002-2004 by the European Commission’s Fifth Framework Programme. **Jane Kaye, Hordur Helgason, Ants Nõmper, Tarmo Sild** and **Lotta Wendel** have written a comparative analysis of the law in Iceland, Estonia, Sweden and the UK. Their paper gives an overview of how the law in

these countries deals with issues such as ownership, consent, feedback, genetic counselling, benefit sharing and access to the database. The article makes it evident how far we still are from a uniform European legal structure for population based genetic databases.

**Antonio Casado da Rocha** explores the ethics of human genetic databases (focusing on the Icelandic one) by making distinctions between models of ownership and inclusion of potential beneficiaries. **Stefan Eriksson** addresses the question of whether results from genetic research should be returned to research subjects and their biological relatives. Although the paper does not raise this question in the context of genetic databases but rather concentrates on the genetic research as such, it allows us to see the problems with feedback that are present also in the settings of genetic databases. This topic is especially relevant for Estonia, since the Estonian Genome Foundation has publicly promised to all participants the possibility to learn of their genetic data and to be informed of their potential or present genetic risks.

The paper written by **Ülle Krikmann, Krista Kruuv, Marju Herodes, Tarmo Sild, Marika Žmenja, Tiina Talvik** and **Arvo Tikk** outlines both the advantages and disadvantages of predictive testing on those with restricted legal capacity by employing practical examples and defining its necessity. The main objective of the article is to find an answer to the question whether minors should be included in the large-scale population-based gene studies.

The other articles in this section highlight the social justice issues surrounding genetic databases. **Sarah Wilson** looks at the specific issues of benefit sharing, ownership and access in relation to three planned database projects (in Iceland, the UK and Estonia). **Minakshi Bhardwaj** asks what genetic databases could mean for developing countries. Although she clearly insists that developing countries should be provided with equal access to the genetic information, she analyses both opportunities and threats that large-scale genetic databases might possess for developing countries.

The paper of **Rainer Kattel** and **Riivo Anton** touches upon questions which are related to both the foundations of economic development as well as to how to sustain development process in catching-up economy for the long term. These issues are discussed, based on the case study of Estonian Genome Foundation Project but those questions are of high relevance also for many other transition societies, both in Central and Eastern Europe and elsewhere. Their paper asks which role the Estonian Genome Project and more generally biotechnology in Estonia might play in future economic development of a country. More specifically they propose what should be done to maximise the chances of success and minimise the risk factors.

**The second part** of the special issue of “Trames” is devoted to the social issues surrounding genetic databases. We learn about public attitudes and different social contexts into which the genetic database projects have been launched.

The question of informed consent of the community comes up in relation to both Icelandic and Estonian genetic databases. The articles of this section help the readers to understand why in some cases the database projects have received overwhelming support whereas elsewhere there has been a lot of discussion or even opposition.

The first article, written by **Külliki Korts**, **Sue Weldon** and **Margrét Lilja Guðmundsdóttir** discusses the public perceptions of population based genetic databases in the three countries where these plans have been most fully developed – UK, Iceland and Estonia. The article introduces the first results of the comparative study carried out in the framework of the European research project ELSAGEN, comparing public attitudes towards science and technology. The authors demonstrate how these perceptions of risks and benefits are rooted in the existence or non-existence of trust towards the institution of science and its governance in different countries.

**Erica Haines** and **Michael Whong-Barr** address ethical issues such as informed consent, altruism and benefit sharing in DNA banking by using the UK-based North Cumbria Community Genetics Project (NCCGP) as a case study. They give valuable insight into the often overlooked aspect in the discussions on the ethical and social aspects of gene databanks, namely the lines of reasoning of the potential donors behind the decision whether to donate or not, which, as pointed out in the paper, are much more ambiguous and context-dependent, often running counter to those generally supposed in theoretical literature.

**Vilhjálmur Árnason** and **Gardar Árnason** analyze the concept of the informed democratic consent, basing it on the example of the debate about the Health Sector Database (HSD) in Iceland. They claim that informed democratic consent has to meet both procedural and substantive criteria and argue that in Iceland the free, reasoned and informed public dialogue which is a necessary condition for such consent, never took place.

The papers of **Sigrídur Thorgeirsdóttir** and **Piia Tammpuu** will allow the reader to compare the public discourses of Iceland and Estonia. They show how national identity is being envisioned and constituted through the national database project, and how the latter is represented and legitimized by appeals to national self-image.

The article of **Sigrídur Thorgeirsdóttir** shows how the image of distinctness of the Icelanders and the purity of the nation is used by the promoters of the database project as a part of the marketing strategy. It points out that the proponents of the database project emphasize the relative genetic homogeneity of the Icelandic population as one of the major assets for a successful and innovative genetic research, whereas other researchers have argued for heterogeneity of the population. The author concludes that these discussions are ideological in nature.

The paper of **Piia Tammpuu** examines the media discourse on the Estonian Genome Project. The study reveals that the Estonian media coverage provides rather contrasting images of the national gene bank. Whereas its initiators and promoters have presented the project as primarily medical and scientific, leading

to better methods of diagnostics and medical treatment and improving the public health-care, critics have regarded the project rather as a business enterprise or questioned the benefits promised by the initiators. The paper shows how in public the EGP has acquired an image of a national project which is closely related to the Estonians' self-image.

The paper of **Tiiu Hallap** asks whether rhetoric or argument has played a more prominent role in the media discussions about the Estonian Genome Project. The article places the observations from the media into the context of some well-known theories concerning science communication. The paper also reflects more generally on the issues of informing public about science and public participation in science communication, considering briefly some recent practical and theoretical developments in science policy in Europe and the US.

The paper of **Külliki Korts** discusses recent survey results of the public attitudes towards genetic research and Estonian Genome Project situating these into the general debate over the impact of gene technology on the modern society, with special attention to the possible social implications accompanying the creation of human genome banks. The paper points out that compared to the Western counterparts, the Estonian population places much more trust in science and scientists. The unchallenged acceptance of gene technology and Estonian Genome Project can at least partly be explained by the fact that there is no tradition of public critical engagement in scientific issues, characteristic to the "reflexive" society, and there is a lack of previous experience in "gene issues".

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