## The new genomics: challenges for ethics

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## 10.1 Summary

## The new genomics: challenges for ethics

Are new concepts needed in biomedical ethics to keep pace with the developments in post-Human Genome Project genomics?
This thesis examines the challenges that developments in post-Human Genome Project (HGP) genomics pose to the established concepts and practices of – Western world – biomedical ethics. These challenges are relevant to the various roles that ethics plays, as for example, its role in the design and the conduct of research, its role in the translation of knowledge and technologies beyond the research stage, and its role as an instrument of governance.
In short, the question here is not about the ways in which ethics influences genomics, as for example by putting constraints on certain applications, but rather the other way round: how does genomics impact on applied ethics and on the role of ethicists. And: what does it mean for the future, given the pace of the developments in the genomics sciences.

### Chapter 1

Chapter 1 provides a broad **Introduction** into the background of the underlying questions and examines how ethics could become a governance tool. It is shown how developments in genetics and genomics coincided with the emergence of a rights-based approach in biomedical ethics, putting emphasis on procedural aspects and on the role that ethics could – or, as assumed for many years: should – play in policy making. This role was reinforced through the institutionalization of ethics in genomics in the form of ELSI and ELSA programs and the establishment of ethics within the Human Genome Organization already in its early years. Chapter 1 also describes the aim of the study, as "to analyze the role of ethics in genomics and the related sciences and to generate hypotheses concerning future developments in ethics, taking into account the likely paradigm shift in the genomic sciences towards systems biology". Concerning the method, reasons are given for taking a hypothesis-generating approach that appears as most appropriate given the dynamics of the developments in genomics. Chapter 1 also includes a short glossary and an overview of the further chapters.

### Chapter 2

**The new genomics: new challenges for ethics?** The article *The new genomics and personal genome information: ethical issues* takes the 'new style' personal genome information as an example to explore the questions that are raised by developments in next-generation sequencing technologies. Starting point is the consideration that "… ethical thinking will inevitably continue to evolve as the science does …" . This supports the claim that current biomedical ethics is no longer fully adequate for addressing the normative issues that arise in the context of post-Human Genome Project Genomics. The advances in sequencing technology are one

example of the very fast developments in genomics where increasing pace and scale require new concepts in large-scale research ethics. Traditional issues of confidentiality and consent are in need of critical review. The global impact of genomics sciences accentuates this need.

### Chapter 3

The chapter **Shifting trends in ethics** discusses in the article *Personalized medicine: new perspectives – new ethics?* the applicability of protection-paradigm based principles to questions arising from the potential conflict between individual and collective interests in population-based genomic research. The development of personalized medicine and, in particular, pharmacogenomics, is taken as an example to demonstrate the need for a new normative framework that comprises new principles like reciprocity, mutuality, solidarity and citizenry, as has been proposed recently. In this context, the concepts of equity and altruism – in particular in the form of health-information altruism deserve scrutiny. Postgenomic medical and research ethics is put in the perspective of a development from traditional medical ethics, clinical research ethics to large scale (genomics) research ethics. It is argued that there is an urgent need for professional ethicists to pro-actively address the new challenges.

### Chapter 4

The chapter **Genomics for all: ethical implications for clinical practice** presents some possible consequences for the physician-patient interaction in view of the new reality of comprehensive personal genome information being available to any individual who wishes to purchase it: *Hippocrates revisited? Old ideals and new realities.* On the other hand, personal information is also being made available to the broad scientific community and to some extent to the public in the new types of studies with open-access databases. This implies giving up (much of) confidentiality and privacy. Once again, this impacts upon consent and requires careful reflection on and renewal of mutual trust in the patient-physician relationship.

### Chapter 5

**New roads to consent. In search of pragmatic moral guidance**. This chapter zooms in on the first demonstration case of this study: The Personal Genome Project. It addresses the concrete question of obtaining valid consent for participation in state-of-the-art genomics research that involves collecting and sharing of comprehensive genotype-phenotype data sets. In *From genetic privacy to open consent* it is argued that 'veracity' should be the primary moral principle guiding novel models of consent. It is argued that taking seriously threats to privacy and confidentiality, in particular in many situations in daily life, leads to the conclusion that promises of privacy and confidentiality cannot realistically be made to participants in biomedical research and, therefore, should not be conditions for consent. Telling the truth about the accessibility of research data will imply asking participants to consent to this degree of

openness. Individuals who are not comfortable with this idea should likely refrain from participation in research. Alternative solutions are scarce.

### Chapter 6

The second case study is presented in Chapter 6, **Current challenges for consent: pharmacogenomics, data sharing, and the language of consent** that consist of two papers showing in which ways current biomedical ethics has reached its limits, taking developments in pharmacogenomics as a model case. In the first paper *A Call for the Creation of Personalized Medicine Databases* we argue for data sharing that includes the vast extant data bases of industry. A moral obligation may exist to use all data that individuals made available for purposes of health research. The ethical challenges arising in the context of data sharing relate to reciprocity, universality and solidarity, and this concerns, among other things, ownership of genomic information, public trust and the requirement of transparency. It remains to be seen, whether the sharing of data between industry and academia can be effectuated by an appeal to 'moral' obligations or that binding regulation (using ethics as instrument for its justification?) will be inevitable.

The second paper in this chapter consists of a working draft of a white paper for the NIH/NIGMS prepared on behalf of the Pharmacogenetics Research Network Data Sharing Action Group: *Pharmacogenetics, data sharing, and biobanking: the scope and limits of consent.*
Designing consent for prospective data collection is less 'difficult' than finding an appropriate way of dealing with extant data (and samples) that are stored in databases and repositories and have been collected long ago – often for other purposes and with widely varying terms of consent. Today, research uses the data from yesterday. But, today we are also generating the extant data of tomorrow. For example in pharmacogenomics research the building of very large well-curated databases and the use of all relevant extant data is indispensable for generating reliable, validated knowledge that will be clinically applicable. A large network like the Pharmacogenetics Research Network has a particular interest in and societal responsibility for making available appropriately consented data.

### Chapter 7

Chapter 7 contains an **Outlook on theory development and education**. The article *Teaching and practicing pharmacogenomics: a complex matter* discusses the importance of the application of systems biology approaches to pharmacogenomics. In particular, attention is paid to the implications of this paradigm shift for the current and future curricula in the biomedical sciences that shall enable implementation of the new approach in clinical practice.

### Chapter 8

Chapter 8 contains the **General Discussion**. This chapter recapitulates the initial questions concerning the role of ethics in genomics and, in particular, the impact upon ethics of the developments in the genomic sciences. The challenges, as posed by these developments are addressed, notably concerning scale and pace, technology and methods, theory development in the sciences up to an evolving paradigm shift, and novel practices. Ethics also faces challenges from 'within': the global science of genomics meets local ethics. Examples of 'particular moralities' – Cofucianism and Buddhism respectively – are presented. Finally, the role is addressed of professional ethicists as involved with science, as 'insiders' or 'outsiders'.

### Epilogue

An **epilogue** on **The $1000 genome** entails a reply to Nature Genetics' Question of the Year in 2007, and discusses the moral desirability of *A just distribution of benefits*.