Ethics and Genetics

Y Joly and BM Knoppers, McGill University, Montreal, QC, Canada

This article is a revision of the previous edition article by O O'Neill, volume 2, pp 659–661, © 2001, Elsevier Inc.

Glossary

Anonymization The process of irrevocably stripping data of identifiers so as to prevent human subject reidentification.
Biobank An organized collection of human biological material and associated information stored for one or more research purposes.
Commodification The transformation of goods and services (or things that may not normally be regarded as goods or services) into a market commodity.
Ethics committee (also called independent review board) An independent group of medical, scientific, and nonscientific members who are responsible for the rights, safety, and well-being of human participants in research projects.
Eugenics Researches or programs that aim to improve the quality of the human race by means of controlled reproduction.
Genetic discrimination The unfair treatment of certain individuals because of actual or perceived interpretations of genetic information.
Genetic exceptionalism The belief that genetic information requires greater protection than other personal or health information because of its presumed special nature.
Genetic (or DNA) profiling The process of determining the distinguishing characteristics of a person's genes for the purpose of identification.
Patent Exclusive rights granted by a government that allows a person to exclude others from making, using, or selling an invention for a limited period of time in exchange for public disclosure of the invention.

Introduction

In the past 20 years, the rapid progression of genetic research has been a source of increasing tension between research ethics in genetics and traditional medical ethics. Evolving from research on single gene disorders to population genomics and now personal genomics, new ethical challenges have emerged. With funding priorities shifting toward translational research initiatives and technology transfer, as well as the advent of commercial direct-to-consumer (DTC) companies, the genetic genie is getting ever closer to health-care management and clinical practice. It is becoming clear that the ethical frameworks developed following the Nuremberg Trials, and other abusive experimentation involving humans, inadequately address the distinct context of twenty-first century genetics. Traditional ethical duties to obtain an informed consent and protect the confidentiality of medical data have taken new contours, while the level of physical risk to research participants is almost nonexistent in genetic research. Adapting ethical guidelines and educating ethics committees (also called independent review board) about the benefits and ethical challenges of contemporary genetic research should be a priority in the coming years.

A realistic look at the ethical issues raised by genetics at the start of the twenty-first century reveals a wide spectrum of possibilities, potential benefits, and ethical issues, as well as numerous efforts to devise policy structures that will ensure that newly acquired genetic knowledge is used ethically. This article will cover some of the more current, recurring issues.

Should Genetic Information Be Treated Differently from Other Medical Information?

With the beginning of the Human Genome Project in the 1990s, genetic information was considered special and therefore to be treated differently from other types of medical information. This current of thought was later coined as 'genetic exceptionalism'. Certain authors claimed that genetic information is a particularly sensitive form of personal information because of its familial and probabilistic nature. They also pointed out that genetic information can be easily stored and shared via Internet databases. Opponents of genetic exceptionalism, however, argued that the issues raised by genetic information are actually quite similar to those raised by other types of sensitive personal or health information. According to these experts, numerous types of health data, quite apart from genetic information, have familial implications and can be just as predictive of future health outcomes (e.g., cholesterol test or HIV status). In their opinion, genetic exceptionalism is a self-fulfilling approach: genetic information is perceived as unique and stigmatizing by community members because genetic-specific legislation and exaggerated coverage in popular and academic media reinforce that view.

Genetics and Privacy

In the coming years, genomic research and personalized medicine will increasingly rely on international data sharing and large-scale biobanking to generate the kind of robust, reliable

 doi:10.1016/B978-0-12-374984-0.00491-5

findings that are needed for regulatory approval and translational research. However, because of recent technological and statistical advances, it has become increasingly difficult to protect the identity of patients and research participants. A small amount of genetic information is now sufficient to reidentify an individual in a genetic database. The value of data anonymization, once thought to be a sufficient safeguard for all genetic privacy issues, has recently been put into question. Solutions proposed to address these new challenges include greater transparency about the limits of applicable privacy protection mechanisms in consent forms; developing elaborate, independent governance structures for biobanks; using state-of-the-art information technology security mechanisms; and moving toward more coherent national and international privacy frameworks. In clinical practice, given the advent of computerized medical files and the fact that a growing number of health workers (e.g., doctors, genetic counselors, nurses, pharmacists, etc.) will have access to genetic information from patients, privacy protection will also become an important factor. Given the popular anxiety generated by risk of confidentiality breaches and misuse of genetic information by third parties (e.g., genetic discrimination), it would be beneficial to gather additional empirical data about the frequency and real impact of these types of events on patients and research participants. Greater knowledge about actual genetic discrimination might both appease unjustified concerns and help better address undesirable scenarios.

Genetics and Medical Care

Given the potential of genetics for health care, its progressive integration into clinical practice should be favorably viewed. The ethical issues arising in clinical genetics are so far quite similar to those arising in other areas of medicine. Typical problems include those of conveying difficult information to patients and their families, including the potential duty to warn at-risk relatives of participants receiving positive test results in specific circumstances and to recontact patients if new meaningful genomic findings surface. As is usually the case in a clinical context, one will need to ensure that genuinely informed consent to tests and treatments is obtained and that the confidentiality of genetic information is protected. Physicians will need to be informed about new genetic clinical tools, medical products, and guidelines and educated on how to properly integrate them into their daily practice. Health-care payers will have to make difficult choices relating to access and reimbursement of new medical devices and therapeutic products, which will likely be expensive. It will also be important to develop thorough legal and ethical frameworks to ensure that genetics is not misused in the clinic to promote eugenic practices or the commodification of human beings, or to provide patients with complex, indecipherable information that simply does not meet clinical utility standards.

Nonmedical Use of Genetic Information

Because of its predictive and identifying properties, genetic information may also be used outside of the medical context. In the insurance and employment fields, the concern is that genetic information could be used by private entities to discriminate and exclude at-risk individuals in order to save costs. A sample scenario could be an insurer requiring insurance candidates to pass a genetic test and refusing life insurance applications from those highly predisposed to developing a condition that would result in early death or offering lower premiums to those presumed to be 'healthy'. As previously discussed, the true extent of genetic discrimination in developed countries remains unknown, but existing data point to a modest incidence. The fact that only a few genetic tests are currently truly useful to predict future health, coupled with the still relatively high cost associated with genetic testing and interpretation of results, will hopefully convince stakeholders to avoid widespread use of genetic discrimination in the near future.

The highly identifying nature of genetic information also explains why it is now increasingly used for criminal enquiries (for profiling (genetic or DNA profiling) and evidence purposes), paternity testing, and immigration proceedings (to verify the existence of a biological link between presumed family members). To prevent abuses and overreliance on genetic data compared to more traditional sources of information (e.g., birth certificates, fingerprints, photographic evidence, oral testimony), clear limits will need to be determined by society and integrated into administrative policies, statutes, and case law.

DTC Genetic Testing

Technical and scientific advancements in genomics have raised expectations among members of the public who would like to see promising research findings translated more quickly into medical practice. Companies, eager to capitalize on this opportunity, have begun offering genetic services directly to consumers via the Internet. They justify their practice by pointing out the need to increase access to valuable new genetic tests and to empower individuals to take charge of their health. However, the practice of DTC genetic testing also raises serious ethical concerns. One of the chief concerns is the questionable validity and clinical utility of many of the tests currently offered. Also troubling is the fact that DTC services are usually provided without adequate genetic counseling, making it difficult for consumers to truly understand the various implications of their results. It should be noted that some of these DTC companies also have commercial research agendas. The high incidence of false or misleading advertising by DTC companies also deserves mention. Efficiently regulating DTC companies is a huge undertaking and a more positive impact might be achieved through the development of simple, easily accessible education programs for the general public.

Population Biobanks

Since 2000, large national population studies that build longitudinal databases and biobanks have emerged. These ongoing studies serve as infrastructures for more specific disease research. Concentrating on the role of the environment on gene expression over time, these resources contain extensive amounts of personal and socio-demographic data, in addition

to blood and urine samples. Since the biobanks can go back to their participants for updates, the data and samples are both historical and contemporary. Participants in such resources derive no personal benefit, are usually asymptomatic (i.e., presumably healthy), and receive no individual results. Overall general results are made available, as well as information on what research has been granted access to the samples and data and for what purpose. A broad consent to future unspecified research is obtained, but in counterbalance, there is a higher degree of data security and ethics oversight and governance. With the advent of whole-genome sequencing technologies that will no doubt be used by researchers accessing the biobank, it remains to be seen whether the 'no return of results' policy will survive, considering that a plethora of health data, including incidental findings (some clinically significant), will become available.

Intellectual Property and Genetics

Early on in the development of genetics, authorities made it clear that nonhuman life forms, otherwise meeting the legal patent requirements, would generally be patentable. More recently, following a few high profile controversies, the application of the patent system to genetic research had to face an increasing amount of criticism. The criticism, based on a number of moral (e.g., the impact of gene patenting on human dignity and humanness), scientific, legal, and economic arguments, eventually convinced authorities to adopt a more restrictive approach to the granting of genetic patents. Today, the thresholds that must be met to obtain genetic patents and the exceptions to patentability vary from one country to another. This creates much uncertainty. For example, patenting an embryonic stem cell is currently not possible in most European countries for reasons of morality, while a recent judgment from a United States Court of Appeals for the Federal Circuit invalidated patent applications on a diagnostic test method to detect hereditary breast cancer. because they claimed unpatentable subject matter. It is presently difficult to assess the economic and social impact of gene patents. However, if genetic research is to proceed with some support from the private sector, reward mechanisms will always remain necessary due to the uncertainties as well as the high cost associated with research and development in this field.

See also: DNA Fingerprinting; DNA Marker; Genetic Diseases; Pharmacogenomics; Population Genetics.

Further Reading

Caulfield T, McGuire AL, Cho M, *et al.* (2008) Research ethics recommendations for whole-genome research: Consensus statement. *PLoS Biology* 6(3): e73.

Howard HC and Borry P (2009) Personal genome testing: Do you know what you are buying? *American Journal of Bioethics* 9(6–7): 11–34.

Kaye J, Heeney C, Hawkins N, de Vries J, and Boddington P (2009) Data sharing in genomics – Re-shaping scientific practice. *Nature Review Genetics* 10: 331–335.

Knoppers BM and Chadwick R (2005) Human genetic research: Emerging trends in ethics. *Nature Reviews Genetics* 6: 75–79.

Rothstein MA (2007) Genetic exceptionalism and legislative pragmatism. *Journal of Law, Medicine & Ethics* 35: 59–65.

Rothstein MA (2010) Is deidentification sufficient to protect health privacy in research? *American Journal of Bioethics* 10(9): 3–11.

Relevant Websites

http://www.coe.int – Council of Europe; Steering Committee on Bioethics (CDBI).
http://www.genomecanada.ca – GenomeCanada; GPS Series.
http://www.humgen.org – HumGen International database.
http://www.nuffieldbioethics.org – Nuffield Council on Bioethics.
http://www.bioethics.gov – Presidential Commission for the Study of Bioethical Issues.