2

Dilemmas of Genetic Information

Urs Dahinden, Nicola Lindsey, Aglaia Chatjouli, Carmen Diego, Björn Fjæstad, Marisa Matias, João Arriscado Nunes and Timo Rusanen

The past decade has seen an increase in public concern about genetics and the life sciences in Europe and the US. Some applications of the technology have barely infiltrated the public arena before they have been rejected, as was the case with genetically modified (GM) foods in much of Europe. Others have been infiltrating slowly, held up by the limits of the technology and our knowledge about genes. Genetic testing is one such area. Despite the great promises of the Human Genome Project to identify the roles of genes and gene sequences in simple and complex diseases and physical traits, the number of clinically relevant genes that can be directly tested for is small. Nevertheless, there is an underlying assumption that comprehensive information about an individual's genetic constitution will be readily available.

We report on public debates surrounding genetic testing and the use of this information. The aim is to investigate some of the properties of genetic information, how it is interpreted in different contexts, and why it may warrant special attention. The text is interspersed with country case studies on particular issues.

What is genetic information?

First we need to define what is meant by *information*, and then *genetic* information. Information is simply a representation of reality by means of symbols (i.e. words, numbers, technical codes and so on). *Genetic* information, then, can be symbolically coded at different levels of abstraction: tests for the presence or absence of a particular allele, tests for deoxyribonucleic acid (DNA) sequences near the gene or genes (genetic markers) and tests for gene products or proteins. Genetic

information can derive from observing a person's physical characteristics, such as their eye and hair colour, or from observing their family members.

Information, therefore, represents genetic reality but is not the gene in and of itself. Information selects and highlights specific features of that reality. This selectivity results from the economic constraints that surround the collection of information. It is a cumbersome process and must be justified by a specific need or purpose. For example, in the clinical context, genetic information is used to diagnose or predict disease.

Information is immaterial, that is, it is not dependent on the continuing existence of the reality it represents. Although information does require some kind of physical storage and management (in the form of texts on paper or electronic data on hard disks), the resources involved are small. Due to its immateriality, information is difficult to destroy. It can be stored at very little cost over a long period, and after the original rationale for collecting it has changed or expired. As a result, information can easily transcend time, space and the social context to which it refers. The immateriality of information also means that it can be efficiently managed – analysing and manipulating symbols requires less effort than managing the physical and social realities themselves.

Information is produced for specific purposes. The process of 'informatization' involves different forms of re-contextualization. Information is gathered and encoded according to the context of collection, it is then stored and transmitted across time, space and social contexts and is finally decoded and re-contextualized by a receiver. As a result, information is multifunctional and can be used for unintended purposes. For example, credit card statements do not just reveal evidence of expenditure but also trace the user in space and time. The possible uses of information are, therefore, multiple and often unexpected.

Modern western societies have often been described as 'information societies' (Bell, 1976, republished in 1999). The debate over the definition of 'information society' and the assessment of its implication remains contested (Castells, 1996; Webster, 2003a, b). Information societies trade tacit and explicit knowledge, and see the development of science and technology as the most important activity. Scientific knowledge and technological products have been seen as the main source of societal and economic change, reshaping societal structures and redefining social values. Freed from the constraints of traditional structures, information societies accelerate fragmentation and individualization. Members of (post-)modern societies do not so much inherit identity by birth and socialization, but rather must construct an identity from a patchwork of social roles and markers.

The emergence of the information society is intimately linked to the globalization of commerce. Genetic information is a new field for business opportunities. This is true for forensic applications as well as medical and pharmaceutical research. The ownership and the access to genetic information is a highly commercial import. Genetic information thus presents a conflict between public and private ownership. The Human Genome Project demonstrates that information that was once hidden and private is being reified and thus made public and a potential commodity. Indeed, the rhetoric of the Human Genome Project

Box 2.1 Case study 1: Patents on nature? The 'Gene Protection Initiative' in Switzerland 2001

Urs Dahinden

The role of patents for the development of biotechnology is controversial. This controversy is by and large an expert debate between a few representatives from industry, public authorities and non-governmental organizations (NGOs). Switzerland is one of the few countries in which a public debate followed by a democratic vote has taken place over this controversy.

What are the main positions in the patenting conflict? On the one hand, the biotechnology industry claims that patents are an important requirement for the commercialization of a new technology. Patents are intellectual property rights that define ownership of scientific and technical information. Patents are the legal instrument by which information can be transformed into a commodity with a private owner and a price. On the other hand, NGOs criticize the current legal practice of patenting biotechnology.

The main source of controversy in patenting is the distinction between discovery and invention. Both invention and discoveries provide new information, but in most legal systems, only inventions can be patented. This distinction is justified as follows: a discovery provides new information about natural laws which are public goods that cannot be privatized and commercialized. By contrast, an invention may be based on natural laws, but adds to those an element of creativity and human culture. At first glance, this differentiation between discovery and invention seems simple and straightforward, but its application in the realm of biotechnology is complex and contested. According to this logic, the discovery of a new gene responsible for a specific disease should not be patentable because it does not include a new invention in the sense of a technology. However, current legal practice allows the patenting of this discovery in the sense that the patent covers those innovations (e.g. therapeutic treatments) that are going to be developed in the future based on current information from the discovery.

The question of what can be patented is decided by national and international patenting offices through a combination of theoretical reasoning and pragmatism. They are in charge of drawing a line between discovery and invention, between public and private information, between open access and commercial restriction. However, patenting offices work in an area of conflicting interest and conflicting parties with unbalanced lobbying power. The industry has a strong interest in gene patenting, whilst NGOs oppose this trend.

The Swiss public debate on patenting followed in most respects the general patterns and argumentation discussed above. The debate was triggered by a popular initiative: 'Gene Protection Initiative' (German 'Genschutz-Initiative') (Bonfadelli et al, 2001). This initiative was launched by environmental NGOs and called for the prohibition of the following three things:

1 the production and sale of GM animals;
2 the release of GM plants and animals;
3 the patenting of GM plants and animals.

The motive of the initiative committee was clearly anti-biotechnology (points 1 and 2) and also against patents for genes (point 3).

In the media debate on the initiative, the issue of patenting was not the main topic. The opponents of biotechnology were making their case of ethics, environmental protection and animal protection, while the supporters of biotechnology emphasized biomedical and economic prospects. In the referendum of 7 June 1998, the Swiss electorate rejected this constitutional amendment with a majority of 66 per cent. A follow-up survey of the voting motives (Hardmeier and Scheiwiller, 1998) showed that the issue of patenting was not the determining factor for either side of the conflict: The ban on patenting was only mentioned by those citizens who were supporting biotechnology; only 2 per cent of them mentioned this as an argument to vote against the initiative (Hardmeier and Scheiwiller, 1998, p11). This is an indication that even in Swiss direct democracy, the issue of patenting remains an expert topic.

emphasizes the interests of the 'public good' that the project will serve, while the 'gold rush' for gene patents puts it back into private hands. The controversy over patenting became a national referendum issue in Switzerland, described in case study 1 (Box 2.1).

Uses of genetic information in the European context

Medical uses

The potential uses of genetic information have been publicly debated across Europe over the past decade, triggered by the rhetoric of progress emanating from the Human Genome Project since 1990. Currently, the most common setting for the production and use of genetic information is the clinic. Genetic testing of consenting adults for disease-related genes, for both diagnostic and predictive purposes, is well supported and fairly widespread across Europe. In most countries, the testing service is offered in nationally approved clinics where adequate counselling can be provided, and not over the counter or through the internet. There has been greater concern about the genetic testing of children, and about prenatal diagnosis and pre-implantation genetic diagnosis. Regulation varies, and some countries have debated more or less pragmatically what traits it should be permissible to test for. These are usually disease related, and exclude diagnosing the sex of an embryo.

Clinical practice is closely related to research. Many mutations are very rare. Patients coming forward for testing for rare mutations may be asked to take part in research studies. However, in order to identify the significance of these mutations in the population, and to develop drug and gene therapies more efficiently, large numbers of participants are needed. Many countries have put forward proposals for large-scale population databases where cohorts of up to a million people can have their lives followed and their diseases monitored and

Box 2.2 Case study 2: Uman Genomics: Biobanking between public and private ownership (Sweden)

Björn Fjæstad

The medical biobank of Västerbotten County Council and Umeå University was founded in 1987. Västerbotten County in northern Sweden has a population of just over 250,000. The county council provides public medical services. At health examinations, patients are asked to donate blood and fill out a questionnaire about health and habits. The biobank consists of some 130,000 samples from 90,000 donors.

In 1999, the stock company Uman Genomics was founded and was granted a monopoly for commercial activities based on the samples (Michael Lövtrup, science journalist, has kindly provided information about the history of Uman Genomics). The product sold was to be knowledge, not blood samples. Up to half of the volume of each sample may be used by Uman Genomics. The privacy protocols and the fact that Uman Genomics had a majority public ownership meant that the design of the operations was seen as ethically exemplary (Abbott, 1999).

The company was not a commercial success. In April 2002, the county council approved a revised contract. Uman Genomics is no longer required to report the results of the analyses to the biobank but can wait until a patent is sought. The requirement for a majority public ownership was revoked.

The legality of this new contract was challenged in court by some biobank employees concerned about who has the right of disposal of the samples and the right to decide over the biobank – the county council or the biobank scientists? The biobank scientists have continuously been encouraged by the county council to seek external funding, which could suggest the biobank is the physical result of a research project and is governed by its main investigators, rather than an integral unit of the county council's normal medical operations. In October 2002, the county administrative court ruled in favour of the county council. This verdict is being appealed against. A higher court will decide whether undue favours were granted to a single business enterprise. Meanwhile the uncertainty in the matter has led to a situation whereby the company is having difficulty finding investors, and in 2003 all employees were given notice.

Another point of conflict involves the conditions of commercialization. The new Swedish biobank law states: 'Tissue samples or parts of tissue samples kept in a biobank must not be transferred or given out for profit purposes'. Some are convinced that the revised contract, fixing a yearly sum, is in agreement with the new law. The critics are just as certain that the opposite is true. In addition, the monopoly granted to Uman Genomics, valid for 20 years, may violate fair competition laws. Some external science funding agencies and the cancer agency of the World Health Organization (IARC) have questioned the formation of Uman Genomics and its use of up to half of the samples in commercial activities.

When Uman Genomics was formed, a new donation document was introduced stating, among other things, that the stock majority is owned by the county council and Umeå University and that transparency is guaranteed. However, the leaders of the county council and the Umeå University have now stated that donors are to be given the opportunity to decide how their blood is to be used.

One of the main founders of the medical biobank has formed a new board for the biobank since they do not accept the new organization created by the county council and Umeå University. The future of Uman Genomics is thus wide open.

compared with their genetic profiles. The best example of this is Iceland, where in 1998 the parliament allowed the whole population's individual health records to be passed on to a large database without patients' explicit prior consent. Individuals are given the opportunity to opt out should they *not* wish to be a part of the study. Access is in the hands of a private company, DeCode. They use this information in conjunction with widely available records of Icelandic family histories and donated genetic material to identify genes involved in simple and complex diseases. Similar databases have been created elsewhere (Maschke, 2005). Governments promote these databases as a valuable economic and national resource.

Sweden and the UK have had such proposals widely debated. Unlike the Icelandic model, these are publicly run and participation is voluntary. Biobank UK, formerly known as the UK Population Biomedical Collection, a database of 500,000 people funded by the Medical Research Council and the Wellcome Trust, has been given government approval. Case study 2 (Box 2.2) reports on the controversy over the commercialization of genetic information in Sweden.

Non-medical uses

Genetic information is also used in non-medical contexts. Genetic fingerprinting, used as person identification, has become an important tool for solving crime in Europe: see case study 3 (Box 2.3). In many countries, massive public and media support have backed high-profile criminal investigations using this technology. This information is also stored in forensic databases, sometimes even if the suspect is not charged or is acquitted, as is the case in the UK. DNA analysis is now used as a routine procedure in paternity testing and has been used in some high-profile paternity suits involving well-known public figures from rock stars to sportsmen. Paternity testing, together with forensic testing, is probably the area where the greatest volume of genetic information has been generated. Despite its relative acceptability, there are still questions about whether the information could be used for other purposes in the future, for example in crime prevention.

Of more serious public concern has been the use of genetic test information by insurance companies and employers. This issue has been debated across Europe, although perhaps not to the same extent as in the US. Currently, legislation prevents insurers using genetic information in Austria, Denmark and Norway, while the Netherlands, France, Sweden and the UK have moratoria in place. The European Council Convention on Human Rights and Biomedicine places a ban on all forms of gene-based discrimination that restricts the use of genetic tests for medical purposes.

Finally, genetic information and the identification of specific genes have raised concern over intellectual property rights. In July 1998, the European directive on

Box 2.3 Case study 3: The forensic use of genetic information in Portugal

João Arriscado Nunes and Marisa Matias

In Portugal, the use of standardized genetic techniques and practices of DNA profiling is now routine in the investigation of criminal cases and paternity claims, and in the identification of victims of disasters.

Scientific and technical standardization, however, is at odds with the specificities of the Portuguese legal system, particularly the admissibility and weight of forensic evidence, and the role of both expert witnesses and judge (Costa and Nunes, 2001). The former are usually forensic scientists working either for the National Institute for Forensic Medicine, for the Laboratory of Scientific Police or for research units licensed for this kind of work. They appear in court as experts of the court, not as experts for the prosecution or the defence. Counter-expertise is rare, although its possibility is inscribed in the law. Judges retain considerable discretionary power in weighing forensic evidence, even if they are compelled to justify its dismissal on scientifically admissible grounds.

The issue of the integrity and quality of the material is critical in so far as this kind of evidence has to withstand scrutiny both as scientific-technical evidence and as legal evidence. The integrity and quality of the biological materials collected in crime scenes is often poor (due to the lack of training in crime scene techniques by police agents). The concept of a chain of custody, as a warrant of the integrity of the evidence, is known in Portuguese legal discourse, but its implementation is contingent on the action of the police, as has been shown by ethnographic research (Costa and Nunes, 2001; Costa et al, 2002; Costa, 2003). Biological material for investigations of paternity claims is collected by medical personnel and its integrity and quality are preserved.

In paternity claims, the use and scrutiny of genetic profiles often takes place as one form of evidence – albeit a crucial one – in a context of conservative conceptions, held by the judge and public prosecutors, of appropriate sexual and procreative behaviour by women and of their moral standing (Machado, 2002). Based on the constitutional right that every citizen has to know the identity of her/his father, Portuguese civil law requires that the public prosecutor's office launch an investigation if the father of a child is unknown. DNA profiling has become the successor technique to blood tests in these investigations.

The creation of genetic databases for forensic purposes has been under discussion over the past two years, but the debate on this issue has been confined mostly to forensic scientists and legal scholars and practitioners.

the legal protection of biotechnological inventions came into force. Essentially, this directive allows genes to be patented provided they have been characterized in isolation of the genome. This has caused debate in Italy, France, Norway, the Netherlands and Germany. Nevertheless, it will be adopted by most European countries. The British debate and the regulation of gene testing for insurance purposes are presented in case study 4 (Box 2.4).

Box 2.4 Case study 4: Genetic testing and insurance in the UK

Nicola Lindsey

The UK has seen three fundamental changes in policy in the past few years on the issue of genetic testing and insurance. In 1997, the insurance industry entered into a voluntary agreement with the government whereby, under the auspices of self-regulation, it would not ask applicants to undergo genetic testing. Moreover, it would only use genetic test information above a certain financial limit. In 1999, the Government decided that if the industry were to be using genetic test information, it ought to be approved officially, and it therefore set up an independent advisory body, the Genetics and Insurance Committee (GAIC), to assess and monitor the industry's use of tests on a case-by-case basis. In September 1999, the committee approved the first test – that for Huntington's disease – to be used in relation to applications for life insurance. The approval triggered a fairly widespread media and public debate and prompted another government advisory body – the Human Genetics Commission (HGC) – to launch a public consultation on the issue. Finding a high degree of public and political concern about the issue, the HGC called for a moratorium on the use of genetic test results just six months later. Finally, in October 2001, faced with growing public pressure the Government conceded to a five-year moratorium, extending the boundaries of the industry's own moratorium in terms of time, financial limits and the range of insurance products covered. It kept the GAIC in place to regulate tests above this limit and monitor the industry's observance of the moratorium.

The two committees were therefore working independently within government and in fact advising the Government on the same issue in opposite ways. The Genetics and Insurance Committee was a technical committee set up to consider the scientific, clinical and actuarial relevance of genetic tests. For this committee, insurance was viewed as a private contract between individual and insurer which is based on the principle of equity – that is, that neither side should be in possession of more relevant information than the other. The evidence that the committee used to assess the relevance of genetic tests was fundamentally quantitative, whereby the insurance company applying for approval must demonstrate that a positive result for the test in question will increase an individual's mortality rate by at least 50 per cent or their morbidity rate by at least 25 per cent. The assumption underlying the existence of the committee is that genetic testing will become more prevalent in the future and therefore it represents an effort to get the regulatory mechanisms in place now.

In contrast, the Human Genetics Commission is a strategic committee set up to consider the social, ethical and legal implications of developments in human genetics. For this committee, life insurance was seen as a social good, that is, something that all individuals should have access to in order to function normally as part of society. In their report on insurance the committee expressed fears that discrimination on genetic grounds by insurance, companies might lead to the exclusion of new social categories and this might therefore discourage individuals from undergoing genetic testing. This was seen as a serious public health issue. In addition, the committee viewed genetic information not simply as the results of genetic tests, but as including family history information. Finally, it argued that genetic tests are not accurate but are always open to interpretation.

Therefore, the two committees may be seen to be operating on different sides of the paradoxes inherent in genetic information and research. The result for policy making in this area in the UK is that the overall regulatory picture is inconsistent and difficult to define.

Regulatory issues

These uses of genetic information raise difficult issues for regulators and policy makers. Genetic information raises questions of consent. For example, is the collection voluntary or coerced? Is informed consent, once given, valid for repeated sampling? How is consent dealt with in cases where the individual is incapable of giving it? Under what circumstances (if any) should individuals be coerced into giving tissue or fluid samples for genetic information? What about forensic settings? Should the sampling of genetic material be compulsory for criminals, suspects or victims of crimes? If the answer is 'yes', what crimes should be included? Should sampling be coercive in cases of paternity claims, and for whom (mother, child, putative father)? Who should be entitled, in each circumstance, to collect the genetic information: medical personnel, research scientists, law enforcement officers? Where should responsibility for authorizing genetic testing lie: with courts of law, medical authorities, research institutions? What are the criteria for inclusion in and exclusion from databases?

Second, questions arise about how genetic information is managed. Who is entitled to manage the information and in what format? Who is in charge of the custody of samples, profiles and of codes for matching individuals with samples and profiles? Should these be kept in separate institutions? What institutional arrangements provide adequate safeguards of privacy? How can intervening institutions and agents be made accountable? What are the mechanisms for controlling violations?

Third, what are the legitimate uses of genetic information and genetic databases? Who is entitled to access the databases, for what purposes, under what circumstances, and how? What are the specific conditions of access for research, medical intervention and forensic uses? Are uses for insurance purposes legitimate? When is it legitimate for employers to use genetic testing for hiring staff and for workplace safety purposes? What safeguards against abusive access are available? Are the threats of misuse or abuse of genetic information different from those that apply to other, more common forms of use of medical or personal information?

Fourth, who owns the genetic information? Is it the person who provided the materials or their family, if the information is relevant for its members or is likely to be sensitive, damaging or a possible source of discrimination for family members (in cases of detection of genetic traits 'running in families', for instance)? Is it the research institute that turned it into manageable and usable data? Can materials or processes based on genetic materials be patented? Who owns the

materials stored in forensic databases? Who is entitled to decide which information to include or exclude from databases?

Discriminatory practices

Finally, how can discriminatory practices based on genetic information be prevented? Are there legitimate forms of discrimination? If so, in which areas and for what purpose? Can individuals be removed from a job on the basis of genetic testing, by invoking the need to safeguard their health and safety? Is it legitimate for an insurance company or an employer to discriminate against non-symptomatic carriers of a genetic trait or a genetic polymorphism ('healthy ill'), invoking increased risk of a disabling disease? How do questions of discrimination relate to current definitions of proportionality and equity in criminal law? Does the inclusion of genetic profiles in databases of convicted criminals violate the rights of those who have been convicted but then released? These and many other questions relating to the safeguarding of citizens against discrimination on the basis of genotypical characteristics need to be examined from regulatory, legal and constitutional points of view. How do such safeguards appear in international and European conventions and how are they then transposed into domestic law?

The above questions reflect the range of complex issues surrounding genetic information with which policy makers in many countries have been grappling. However, in many ways questions relating to genetic information are questions of information in general. The aim of the following sections is to identify the properties of genetic information that render it a particular challenge.

Particularities of genetic information

Genetic information is pervasive and has relevance on many different levels. It has the unique ability to define both individuality and membership of a social group. Individuals construct their identities with reference to membership and non-membership of social groupings or 'microcultures', hence building a dichotomous concept of togetherness and belonging and of otherness. Such typical reference systems include those of kinship, age, gender, ethnicity, economic status and profession. Genetic information may act to reinforce these social groupings, which are also conceptual categories within which the self is constructed, either by adding value to the existing connecting threads that bond the group, or by devaluing them, resulting in the impoverishment of the sense of belonging. In other cases, genetic information could also be constructing new connecting threads, leading to the formation of novel collective structures in formal or informal forms, whether institutionalized or not. Hence, genetic information has the potential power to change the boundaries between the self and the other.

According to Martin (1987), the interpretation process of genetic information by non-experts is not random; it varies according to social status and often reveals other aspects of people's lives. In addition, genetic information and the interpretation of it may lead to the construction of new social communities based on

Box 2.5 Case study 5: The role of genetic information in reproduction among same-sex couples in Finland

Timo Rusanen

In Finland, same-sex unions have been legal since 2002 as marriage-like registered partnerships giving many of the legal rights typical for married heterosexual couples. Whether or not same-sex couples should have the right to adopt children is still being debated. More pertinent to this debate is that the existence of fertility treatments using artificial insemination opens up the possibility of biological parenthood for one of the partners in a same-sex couple.

The role of genetic information is highly relevant for biological parenthood, where the homosexual couple have to make a decision about whose eggs or sperm of the two partners will be used for the treatment. In addition, a decision has to be made about the origin of the eggs or sperm to be used as a complement from the donation bank. No matter what selection criteria the couple may apply, these decisions rely heavily on genetic information about the potential parents and about the egg or sperm donors. In sum, the artificial insemination technique offers to homosexual, and incidentally to heterosexual, couples the possibility of a biological parenthood that can be designed, based on genetic information.

Finland's Health Care Ethical Committee (ETENE) allows fertility treatment only to heterosexual couples. Treatment for single women, but not for homosexual couples, is allowed. One reason for this is that the current bill on the registration rights of same-sex couples does not allow adoption (HS 2001). The Ethical Committee allows fertility treatments to heterosexuals only on the basis that the position of the child is paramount. However, there is also a view that prohibiting same-sex couples from acquiring joint children would be a form of discrimination against sexual minorities. Furthermore, if homosexual couples had access to fertility treatment, this would be inconsistent with the situation where same-sex couples have no right to adopt each other's children. Under the present Finnish legislation, if one partner in the homosexual couple has a biological child, the other partner cannot adopt it. In this respect, allowing infertility treatment without the possibility of adoption would be illogical in the context of the existing legislation (www.etene.org).

shared genetic traits. Genetic information has the potential to redefine the social in biological terms. Rapid advances in the production of genetic knowledge bring the 'biological' into sharp focus. This process has become known as 'geneticization'. Geneticization is a reductionist process that places the organic, the mechanistic and the biological as the basis of human existence while devaluing social experiences and feelings (Lippman, 1992; Nelkin and Lindee, 1995). According to this geneticization trend, the self, the personality and the potential of the individual are genetically determined. The relationship between the 'social' and the 'biological' has historically been a source of controversy and hence the evident power of genetic information to reorganize this relationship explains, in part at least, its controversial nature.

Genes and personal identity

This redefinition of the social order creates new dilemmas that may threaten an individual's sense of autonomy. This can be witnessed, for example, by the emergence of people born through sperm and egg donation who now want to trace their biological fathers and mothers. In addition, new reproduction technologies open the possibility of biological parenthood to same sex couples. Case study 5 (Box 2.5) sketches some of the questions and conflicts regarding the role of genetic information in reproduction among same-sex couples in Finland.

Reproduction technologies are changing our understanding of what it means to be a parent. Individuals who undergo genetic testing are simultaneously implicating members of their own family in both present and future generations. Some family members may not want to know they are at risk. Those who have tests have to decide whether and how to act on the information – should someone who knows that they carry the Huntington's gene go ahead and have children of their own, knowing they have a 50 per cent chance of passing it on? Should they actively screen out embryos that carry that gene?

Discourses and social representations of genetic information highlight the multiplicity of meanings that can be given to this information in different socio-cultural contexts, beyond, or apart from, its scientific-biological meaning. Genetic information brings novel meanings and at the same time strengthens old meanings, symbols and myths all of which make up the material used in the process of constructing one's identity and the boundaries that separate the 'self' from the 'other', 'us' from 'them'.

Redefinition of kinship, disease and ethnicity

The three most obvious areas where genetic information can be seen to redefine social groupings are the family, the disease group and the ethnic group.

Family members are biologically related, that is, they share inherited biological substance. In addition they are connected by emotional feelings and economic relationships. The strength of these connections may differ in different cultures. In the west, for instance, biological relatedness has been the prevalent determinant. In Euro-American kinship, blood ties and the sharing of bio-genetic substance make up central organizing symbols in lay concepts about kinship. The idiom of nature is central in the American kinship system. 'The family is formed according to the laws of nature and exists according to laws that are experienced by the people as natural' (Schneider, 1980, p34).

However, in many cultures the extended family form has been devalued. For example, second cousins are often not felt to be relatives. In such contexts, the appearance of genetic information may emphasize the biological connections (geneticization). Once a family member is informed of an inherited genetic condition, he/she will be challenged to inform the other family members and to trace the specific genotype in the family tree. In this example, the family as a group is then defined via its biological/genetic characteristics.

People who have a specific genetic disease in common may form social groupings. In this case, genetic information might be the cause of the formation of the grouping, which enables the members to adopt common ways of dealing with the problem they share. They might evolve a collective discourse that will represent them in society and will have an economic, political and emotional impact. We are witnessing the emergence and empowerment of such groups that elaborate political and ideological discourses regarding their 'special' conditions and use the genetic information in multiple ways. For example, some groups via their collective actions argue that their unique genetic profiles should not be stigmatized and, drawing on their human rights, they may embrace their genetic difference rather than trying to change it with future gene therapies (the case of congenitally deaf people is often quoted in this context). Other groups try via their collective actions to demand attention from the various stakeholders, and ask for special treatment, in the workplace or in economic support. In many cases, they ask the scientists to give priority to their problem, and to drive research towards a direction best suited to their specific problem.

The progress of genetics has revealed information regarding the differences and similarities of the genetic profiles between and within ethnic groups. This genetic information has mainly been produced by the Human Genome Diversity Programme and has begun to play an important role in the process of the construction of ethnic identities. Ethnic groups may use the information in multiple ways, and may allocate different meanings to it. For instance, such information may reconstruct the foundations of an ethnic group, in terms of the determinants that make members of an ethnic group different from the rest. Alternatively, ethnic groups may use this information on a political level, in order to claim their difference, or even to claim separate land, extra rights and so on. For example, there is evidence to suggest that native Australian Aborigines are adopting genetic testing technologies in deciding their new political representatives.

The multifunctionality of genetic information

Human genetic information is comprehensive and therefore multifunctional. Our current scientific understanding of human genetics is limited. Even though it is known that many human properties are determined to a greater or lesser extent by genetic predispositions, the determining influence of other factors such as environment or individual choice is not known. While there is scientific and political controversy about this topic, it is illuminating that in this discussion on heredity virtually every human property is addressed, ranging from physical (e.g. body size) to social (e.g. criminality), psychological (e.g. mental health, homosexuality) and intellectual characteristics (e.g. intelligence). It is this comprehensiveness that enables genetic information to be used for very different purposes: for identifying a person or for determining his or her predisposition for specific illnesses. This multifunctionality of genetic information presents a key challenge for data protection and privacy. Genetic information that has been gathered for diagnostic purposes might potentially also be of interest for insurers, employers and public health administrations. However, the norm of privacy states that individuals should be in control of their personal information.

We are witnessing in western countries via the symbolic use of the 'genetic' a 'new cartography of the body redefining health, disease and fault in a realistic, biologically deterministic way' (Strathern, 1992). This phenomenon is part of a wider turn according to which inheritance and nature are being reproduced as techniques (Rabinow, 1996) and blood kinship is being geneticized, medicalized and instrumentalized.

Power and hierarchy: The lay–expert divide

The genetic testing procedure is unlike most other medical testing procedures in that the patient not only receives medical or technical advice but also undergoes intensive counselling from a trained genetic counsellor. This need for counselling is driven by the complexity and sensitivity of the information rather than the physical pain associated with the procedure itself, which is minimal. A hierarchical divide and possibly a power differential are thus created between those who possess the information and those who are authorized to access and interpret it. As the importance of genetic testing grows, this divide will inevitably grow wider.

Sociological researchers have shown how the development of genetic testing and the focus on the genetic basis of disease have led to a reclassification or 'geneticization' of disease diagnoses in the clinic. So, those who have non-familial breast cancer are treated differently from those who have inherited forms, despite having virtually the same clinical symptoms. But more seriously, as genes involved in more complex traits are identified, a process of medicalization is taking place where the boundaries of what is normal are constantly being shifted with the identification of each new gene and mutated allele. The label 'medicalization' summarizes the trend, that more and more societal problems are defined in terms of medical categories. Examples can be found in other policy fields, for example in relation to drugs, crime and the labour market. The results of pre-symptomatic tests challenge the current social definition of health and illness: having a positive test result without symptoms of a disease does not fit into either category. Behavioural traits, such as aggression, sexuality and alcoholism, are in the process of being redefined in medical terms. With the power of access to our genes lying in scientists' hands, will the result be that normality becomes a medical condition? Will we be turning to scientists to define who we are? Or worse, will large social institutions like insurance companies be able to find out things about us before we even know them ourselves?

The twin processes of medicalization and geneticization challenge a traditional understanding of how we view disease and the inheritance of particular traits. Although the genetic definition of a disease may be new, beliefs about inheritance are not. They have long been part of social and family cultures, for example through the identification of family resemblances. They may also be associated with social and cultural practices concerning the inheritance of wealth and personal possessions. These beliefs are likely to affect how people of different cultures react to genetic information in the clinic. For example, clinicians have

reported how patients with inherited forms of breast cancer fail to consider that the gene may have been inherited through the paternal line because they assume it can only be passed on by women. These beliefs cannot be ignored, because they influence how information is processed and understood by patients. Similarly, the experts' understanding of inheritance and the context in which they work will influence the conception of the genetic testing procedures and the destiny of the results. Factors concerning the management and ownership of the testing will also influence the relationship between patient and medical expert. Therefore, given the complexity of the genetic testing and the sensitivity of the information, these procedures require not only medical, but also psychological and sociological expertise, usually provided in the form of genetic counselling both before and after testing.

While accepting that the way in which scientific and technological knowledge circulates is influenced by the expert perspective (insiders) on sharing knowledge with lay people (outsiders) (Bauer and Gaskell, 1999), it should also be recognized that lay–expert hierarchies are not simple power relations defined over who does know (experts) and who does not know (lay) about genetic testing. The complexity of the issue and the constraints of our knowledge affect all the people involved, with the result that experts in some fields are at the same time lay people in others. Therefore, the separation between lay and expert knowledge is not a simple one (Moscovici, 1981) and we must consider several types of expertise and lay knowledge (Wynne, 1995, 1996; Irwin and Wynne, 1996). As an alternative to the so-called deficit model of the public understanding of science, Wynne (1995, 1996) and Irwin and Wynne (1996) suggest a process of 'creative construction' in which expert knowledge is transformed into lay knowledge, where the representations of science are seen as a common-sense answer to the challenges made by science and scientists in present-day societies.

Thus, the relations between lay people and experts are deeply related to the contexts, norms and rules of testing procedures as well as to the understandings of cultural inheritance on the part of both groups.

Limitations of genetic prediction and intergenerational responsibility

Cultural understandings of inheritance, family and social relationships add a further layer of interpretive complexity to genetic information. At the same time, genetic information is inherently probabilistic information. On its own, genetic information cannot predict when a disease will develop, what the symptoms will be or how severe it will become. Genetic tests can never be 100 per cent predictive because the same disorder can be caused by many different gene mutations and can arise spontaneously in the population. Lifestyle choices or prophylactic treatments may help to combat some genetic diseases, but it is uncertain what the effect of these and other environmental influences will be on any given individual.

Therefore, genetic information – the quantitative estimate of disease – is virtually useless on its own; it must always be accompanied by an interpretation

or a qualitative element. But difficult decisions often rest on these interpretations, such as whether to go ahead with a pregnancy or whether to inform existing children and siblings of their risk. The meaning of genetic risk is therefore more global than just an index or statistical statement can provide. Not only are the risk estimates intrinsically difficult to understand in real terms, but they are also affected by the seriousness of decisions that must be based on them. These decisions will be laden with fundamental social and personal values as well as psychological factors and, as a result, negative and positive outcomes may be attributed with greater significance than is predicted by their statistical risk alone. At the same time, clinically, the information is often of little value at all if tests become available before any preventive treatment or therapy is developed. It is not surprising then that a large proportion of people at risk of Huntington's disease – a highly penetrant neurological disorder of later life – choose not to undergo genetic testing.

A further implication of this is that the new production of genetic information is intimately linked with developments in information technology. On the one hand, the success of modern human genetics would not have been possible without large computers and robots doing much of the analysis in an automatic, reliable and fast way. There is indeed a new academic discipline emerging from this successful cooperation, called bio-informatics. On the other hand, modern computer technology is also making some of the problems related to the abuse of genetic information worse, due to the size of genetic databases and the speed at which they can be accessed, not to mention the restrictions over who is able to access it.

Finally, genetic information is also fundamentally temporal information. It is diagnostic and predictive. Genetic information has implications not only for the individual concerned but also, crucially, for future generations. Through the use of family history, it draws past, present and future generations into a single experience. However, it is the history of past uses of genetic information that causes people to fear the future. One need only look at the eugenic policies of the 1930s and 1940s to know how risky the use of genetic information for some people might be. There is a fear among many people that the use of genetic information to select desired traits or characteristics and to penalize those without is a 'slippery slope'; once going down that route, there is no breaking the trend. There is a sense that genetic information is irreversible – once we know, there is no way of ignoring it. We have to carry the burden of that knowledge with us throughout our lives. It is no surprise that people who are found to carry a disease-linked gene perceive the positive test result as the first discernible symptoms of that disease – even if the gene is recessive and they are not actually at risk of developing symptoms. The enlightenment imperative of 'dare to know' might appear in a new light: knowledge as an unwanted curse or burden.

Some paradoxes by way of conclusion

In this chapter, we map out some of the features of genetic information that define its 'special' status. In doing so, a number of paradoxes have been uncovered. Genetic information can be both *concrete* and *abstract*. We cannot experience a gene, only its effects. Yet to identify a 'gene' is to reify a physical property present in every cell of the body. Genetic information is both *simple* and *complex*. It is simple to obtain and can be reduced to a single yes/no presence or absence of an allele, but at the same time it can only have relevance within a system – the environment of other genes, the cell, the body, nature and so on. Genetic information is often presented statistically, while its social *meaning is not quantifiable*. While we do not have the technology to treat genetic diseases, is it worth knowing that we have a higher chance of developing a problem? At the same time, genes interact in a system with each other and with the environment. Is it worth knowing about one gene if it can be mitigated by another about which we do not know? Genetic information has the power to define both *individuals* and *groups*. It is inherently structural information. It can redefine social relationships and the social order. Genetic information can show *unity* or *diversity*, depending on the focus of public discourse. On the one hand, genes are something that belong to all of us – the human genome – and are therefore a public good of humanity as a whole. On the other hand, an individual's genome is also personal and should therefore be in the control of that private person. So, genetic information has the power to make the public private and the private public. It is something that might be traded as a private good while used for the public good. Genetic information is *multifunctional*. It can describe medical and non-medical traits and be used for medical and non-medical purposes. Genetic information is often thought of as new because it requires the application of new and better technology. But we have long been aware of our physical differences, of how these are passed on in families and how they may be associated with different social groups. Genetic information is therefore not new, but a redefinition of old information in new terms. This process of valuing genetic information is often referred to as 'geneticization'. Some genetic information is not particularly sensitive, such as blood groups, gender, fingerprinting for crime. In other contexts, it is extremely sensitive, such as carrier status of a genetic disorder.

These paradoxes highlight the ambivalent nature of genetic information. Similar to the development of other technologies, one would expect that this ambivalence means a temporary openness that will close in the social shaping of technology. What will be the societal impact of genetic information? Will the current trend of geneticization in medicine and childbearing prevail? It is premature to provide definite answers, because conflicting tendencies coexist. On the one hand, genetic information is a complex issue debated only within a small expert community. On the other hand, our case studies showed public sensitivity towards this topic in many European countries, not least because the very definition of personal identity is at stake. The future of genetic information is wide open.

It is the enduring belief in the potential of genes that causes the debate around the use of genetic research to persist. Therefore, any usage of genetic information

in individual and political decisions should proceed cautiously. The certainties of today might prove to be the errors of tomorrow. On the other hand, scientific progress is a strong legitimization for current efforts in the collection and analysis of genetic information. As the Iceland example shows, a popular pastime of family interests in genealogy has become a valuable resource for modern medical research. The question arises: if at all, then to what extent does anticipated future progress justify the partial suspension of fundamental rights (e.g. protection of privacy and personal data) at the present time.

References

Abbott, A. (1999) 'Sweden sets ethical standards for use of genetic "biobanks"', *Nature*, vol 400, no 6739, p3

Bauer, M. and Gaskell, G. (1999) 'Towards a paradigm for research on social representations', *Journal for the Theory of Social Behaviour*, vol 29, pp163–185

Bell, D. (1976, republished in 1999) *The Coming of Post-Industrial Society: A Venture in Social Forecasting*, Basic Books, New York

Bonfadelli, H., Dahinden, U., Leonarz, M., Schanne, M., Schneider, C. and Knickenberg, S. (2001) 'Biotechnology in Switzerland: From street demonstrations to regulations, in Gaskell, G. and Bauer, M. W. (eds) *Biotechnology 1996–2000: The Years of Controversy*, Science Museum, London, pp282–291

Castells, M. (1996) *The Information Age: Economy, Society and Culture*, Blackwell Publishers, Malden, MA

Costa, S. (2003) *A justiça em laboratório: A identificação por perfis genéticos de ADN entre a harmonização transnacional e a apropriação local*, Almedina, Coimbra

Costa, S., Machado, H. and Nunes, J. A. (2002) 'O ADN e a justiça: A biologia forense e o direito como mediadores entre a ciência e os cidadãos', in Gonçalves, M. E. (ed) *Os Portugueses e a ciência*, Publicações Dom Quixote/Observatório das Ciências e das Tecnologias, Lisbon, pp199–233

Costa, S. and Nunes, J. A. (2001) 'As atribulações da ciência 'impura': A harmonização da biologia forense e a diversidade dos sistemas jurídicos', in Nunes, J. A. and Gonçalves, M. E. (eds) *Enteados de Galileu? Semiperiferia e Intermediação no Sistema Mundial da Ciência*, Afrontamento, Oporto, pp107–141

ETENE (2004) Homepage of the Finnish National Advisory Board on Health Care Ethics (ETENE), www.etene.org

Hardmeier, S. and Scheiwiller, D. (1998) 'Analyse der eidgenössischen Abstimmungen vom 7 Juni 1998: Volksinitiative "Zum Schutz von Leben und Umwelt vor Genmanipulation" (Genschutz-Initiative)', *Vox-Analysen eidgenössischer Urnengänge*, vol 63, GfS-Forschungsinstitut, Bern, Institut für Politikwissenschaft der Universität Zürich, Zürich

HS (2001) 'Ethical committee would allow fertility treatments only to heterosexuals', *Helsingin Sanomat*, 1 February

Irwin, A. and Wynne, W. (1996) *Misunderstanding Science? The Public Reconstruction of Science and Technology*, Cambridge University Press, Cambridge

Lippman, A. (1992) 'Led (astray) by genetic maps: The cartography of the Human Genome Project and health care', *Social Science and Medicine*, vol 35, no 12

Machado, H. (2002) 'Tribunais, género, ciência e cidadania uma abordagem sociológica da investigação judicial de paternidade', Doctoral dissertation in Sociology, Universidade do Minho, Braga

Martin, E. (1987) *The Women in the Body: A Cultural Analysis of Reproduction*, Beacon Press, Boston

Maschke, K. J. (2005) 'Navigating an ethical patchwork – Human gene banks', *Nature Biotechnology*, vol 23, no 5, pp539–545

Moscovici, S. (1981) 'On social representations', in Forgas, J. P. (ed) *Social Cognition: Perspectives on Everyday Understanding*, Academic Press, London, pp181–209

Nelkin, D. and Lindee, S. (1995) *The DNA Mystique: The Gene as a Cultural Icon*, W. H. Freeman, New York

Rabinow, P. (1996) *Essays on the Anthropology of Reason*, Princeton University Press, Princeton, NJ

Schneider, D. M. (1980) *American Kinship: A Cultural Account*, 2nd edn, University of Chicago Press, Chicago

Strathern, M. (1992) *After Nature: English Kinship in the Late Twentieth Century*, Cambridge University Press, Cambridge

Webster, F. (2003a) *Theories of the Information Society* (International Library of Sociology), Routledge, London and New York

Webster, F. (ed) (2003b) *The Information Society Reader*, Routledge, London and New York

Wynne, B. (1995) 'Public understanding of science', in Jasanoff, S., Markle, G., Pinch, T. and Petersen, J. (eds) *Handbook of Science and Technology Studies*, Sage, London

Wynne, B. (1996) 'May the sheep safely graze? A reflexive view of the expert–lay knowledge divide', in Lash, S., Szerszynski, B. and Wynne, B. (eds) *Risk, Environment and Modernity: Towards a New Ecology*, Sage, London, pp44–83