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Human Genetic Data: Preliminary Study by the IBC on its Collection, Processing, Storage and Use

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This Report was revised on the basis of discussions at the Eighth Session of the IBC (Paris, 12-14 September 2001) and of written comments sent by members of the IBC. It was further revised on the basis of discussions at the first meeting of the Drafting Group of the IBC (Paris, 11-12 February 2002) and written comments sent by members of that group.

The purpose of the Report is to document the range of issues contained within the topic "Collection, Processing, Storage and Use of Human Genetic Data". It is by nature a preliminary document and should not be regarded as definitive. It is recognized that some issues need further development, for example issues concerning databases, ownership of human genetic data, the differing requirements of identified, de-identified and anonymous biological samples and the impact of the purpose (medical, research, forensic, etc.) for which the samples were collected on the procedures. The guidelines themselves, including both their order and number, are to be regarded as preliminary.

As agreed at the Eighth Session, this Report will be used by the Drafting Group of the IBC as a working document for the drawing up of an international instrument on human genetic data.

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I. INTRODUCTION

1. Every scientific revolution brings with it a host of ethical and social questions. The so-called genetics revolution is no exception, giving rise to a broad international debate on how the undoubted benefits of progress in this area can be reconciled with certain core human values.

2. General guidance for researchers concerning the conduct of research with human participants has been provided in various international documents, particularly the Nuremberg Code, the World Medical Association Declaration of Helsinki and the International Ethical Guidelines for Biomedical Research involving Human Subjects of the Council for International Organisations of Medical Sciences (CIOMS).

3. One of the most significant contributions to this debate has been UNESCO's *Universal Declaration on the Human Genome and Human Rights*, adopted by the General Conference of UNESCO in 1997 and endorsed by the General Assembly of the United Nations in 1998. This document, which was conceived and elaborated by the International Bioethics Committee of UNESCO (IBC), has proved to be a firm foundation for the debate, setting out basic propositions as to the moral status of the human genome. It represents international consensus on how our genetic inheritance should be treated, placing moral limits to what may be done with genetic knowledge.

4. The *Universal Declaration on the Human Genome and Human Rights* has proved to be a good reference point in a complex and sometimes confusing debate. Many voices have been raised, and there have been numerous statements on ethical issues by governmental and intergovernmental sources. Science itself has not ignored its responsibilities in this area, and throughout the world scientific organisations have commented on the ethical principles which should guide the direction of scientific endeavour in genetics. The *Declaration on Science and the Use of Scientific Knowledge*, which was adopted at the UNESCO/ICSU World Conference on Science and endorsed by the General Conference of UNESCO in 1999, also provides guidance. As a result, a body of recommendations and regulations has built up.

5. The IBC has specific responsibility for promoting the *Universal Declaration on the Human Genome and Human Rights* and explaining its implications, a matter which it reported upon in its Report on this issue (*Report of the Working Group on the Follow-up of the Universal Declaration on the Human Genome and Human Rights*, 1999). The follow-up work has continued, too, in a number of other forms. In particular, a working group was set up to investigate issues of confidentiality, and this resulted in the publication of the IBC's *Report on Confidentiality and Genetic Data*⁽¹⁾.

6. While there are indications that the *Universal Declaration on the Human Genome and Human Rights* is reaching its desired audience and is being taken into account in national deliberations, the need for UNESCO to play a role in the international debate on the ethics of genetics continues. Indeed, given the rapid developments in human genetics with increasing numbers of genetic data banks being established, controversial uses of genetic data, and increasing non-medical use, it could be argued that this role is now more important than ever and one which UNESCO is uniquely placed to fulfil. Through the encouragement of debate, UNESCO can assist individuals, institutions and States to translate the spirit of the *Universal Declaration on the Human Genome and Human Rights* into more concrete areas of concern.

1. These reports have been published in the *Proceedings of the Sixth Session of the IBC* (Rabat, Morocco, 1999) and are available on Internet (www.unesco.org/ethics).

7. At its meeting in May 2001 the Intergovernmental Bioethics Committee (IGBC) of UNESCO made the recommendation that the IBC should, when examining the issues related to genetic data, “distinguish between the different types of data and continue to examine the issue of disclosure of genetic information to third parties”.

8. The Director-General of UNESCO has, therefore, identified the issue of human genetic data as an area in which the IBC might assist in the elaboration of an instrument, addressed to the international community, setting out the principles under which human genetic data should be handled, and this recommendation is contained within the Programme and Budget for 2002-2003 (31 C/5), which was approved by the General Conference of UNESCO in November 2001. The precise form of this instrument remains to be settled, but it has been proposed that ultimately there is a place for a declaration of the same status as the existing *Universal Declaration on the Human Genome and Human Rights*, sitting alongside it and complementing the principles which it proclaims.

9. The project would not be concerned with some of the broader ethical issues which are dealt with in the *Universal Declaration on the Human Genome and Human Rights*. Its focus would be on the very specific issue of how genetic information about individual persons, families and populations is collected, processed, stored and used. While this area may seem narrow in scope, it gives rise to major questions of principle. It is also a central point in the wider debate on how personal information is to be protected in a world in which such information may be gathered and distributed with ever-increasing ease. It therefore addresses fundamental issues of human dignity as well as touching upon the fears which many people have as to how knowledge of some of the most intimate details of their lives might be protected from abuse. The topic is therefore one of considerable human rights significance while at the same time being one of major concern to the public.

10. In order to explore this issue a Working Group of the IBC was set up and met at UNESCO Headquarters on 14 and 15 June 2001 to discuss the possibility of drafting such an instrument (see Composition of the Working Group in Annex). During the course of the meeting, a wide range of issues was examined, beginning with a consideration of whether such an instrument was needed and ending with an attempt to identify, on a preliminary basis, what matters might be included within the instrument.

II. THE NEED FOR AN INSTRUMENT

11. While numerous reports, recommendations, codes and laws dealing with the issue of human genetic data are in existence, the *Universal Declaration on the Human Genome and Human Rights* is currently the only universal international instrument.

12. At a regional international level, a prominent instrument is the Council of Europe's *Convention on Human Rights and Biomedicine*, several articles of which are directly or indirectly concerned with genetic information. (Work is currently proceeding on the drafting of a genetics protocol to this Convention.)

13. Also at a regional international level, but emanating from non-governmental organisations, are ethical statements from bodies such as the World Medical Association (Declaration on the Human Genome Project, 1992) and the Ethical Legal and Social Issues Committee of the Human Genome Organization (Statement on DNA Sampling Control and Access, 1998).

14. At a national level, statements and codes have been issued by research funding organisations, national ethical committees or professional bodies. Examples include statements from the Council for Science and Technology of Japan (*Fundamental Principles of Research on the Human Genome*, 2000), the American College of Medical Genetics (*Statement on Storage and Use of Genetics*, 1995), the UK Clinical Genetics Society (*Guidelines for DNA Banking*, 1989) and the Health Research Council of New Zealand (*Ethical Considerations for Research in Human Genetics*, 1998). There are also national data protection or privacy laws, the effect of which is to establish protection, to a greater or lesser extent, of the confidentiality of human genetic data.

15. Even though there are already many statements, guidelines and codes dealing with the issue of genetic data, the changing conditions in which genetic research is being conducted creates a strong case for a universal international instrument. These changing conditions include the increasing involvement of the private sector, the rapid increase in the number of human genetic databases, the controversial nature of some proposed uses and the international character of genetic research, this latter factor introducing the possibility of variation in the standards applied to research and variation in the regulatory frameworks. In particular, researchers may seek raw genetic data from people living in countries where there may not be an adequate level of protection. The need to recognize the rights of vulnerable populations in respect of genetic data is crucial, and the authorities in such countries may be expected to welcome international guidance on appropriate standards of protection. By virtue of its acknowledged status as guardian of cultural and intellectual patrimonies, UNESCO is better placed to provide this guidance than most other bodies.

III. DEFINITION OF HUMAN GENETIC DATA

16. The human body is made up of a large number of cells with similar characteristics. Each human cell contains a nucleus that contains 23 pairs of chromosomes. One member of each pair of chromosomes in an individual is derived from the father and the other from the mother. Each chromosome contains DNA which carries genetic information in an encoded form. All the DNA contained in all the chromosomes is called the genome. Biological samples from which DNA is commonly extracted are blood, tissue, cells from the inside of the mouth or other body fluids such as semen. Blood samples may include cord blood which is representative of the foetus.

17. A gene is a segment of DNA that contains information for the synthesis of RNA molecules required for synthesis of proteins within the body. The human genome is believed to contain of the order of 30,000 genes, and a complete set of genes is present in every cell of the human body. Genes are discontinuous and include non-coding regions as well as regions coding for proteins. Presently the functions, if any, of most non-coding regions are not known.

18. Genetic information is encoded in the DNA as a sequence of nucleotides. This information is passed on from one generation to the next, virtually unmodified. The sequence of nucleotides in the DNA may be determined and stored, for example in a computerized DNA data bank.

19. Thus, genetic material and genetic information (the sequence of nucleotides in the DNA) can be separated, and the information can be stored separately from the biological material from which it was determined and thus become available for subsequent research. As such genetic information acquires a status of its own.

20. DNA sequences between two individuals are largely similar, but contain a sufficient number of differences to be able to distinguish them based on their DNA sequences. Often, certain characteristics of the DNA sequences are exploited for easier DNA profiling of individuals. One of these characteristics is that segments, typically non-coding segments, of the DNA contain short sequences of nucleotides that are randomly repeated a large number of times. Often there is variation in the number of repeats across individuals. The number of repeats at many such highly-variable DNA segments provides the DNA profile of an individual. Such DNA profiling data are commonly used in crime detection and forensic medicine and in some countries there are now large databases of DNA profiles.

21. Information about the number and state of the chromosomes, called a karyotype, is also relevant to a person's genetic identity and may be determined from a laboratory examination of a blood sample. Abnormalities in chromosome structure are generally not conserved through generations. The written description of the karyotype thus constitutes another form of human genetic data. The slides used in the laboratory for karyotype examination may also be stored and therefore also need to be considered.

22. The term human genetic data thus includes karyotype data, DNA sequences, DNA sequence variants (called alleles), such as mutations, single nucleotide polymorphisms, short tandem repeat polymorphisms and insertion/deletion polymorphisms. While this Report is focused on the issues of collection, processing, storage and use of human genetic data, it will also include issues concerning the collection, processing, storage and use of the biological samples from which the human genetic data are derived.

23. In a broader sense, the term human genetic data, when applied to an individual, may be taken to include any information about the operation of heredity in the case of that person. This information may be derived in a number of ways.

- (a) *The taking of a family history through interviews with family members and the analysis of medical records supplied by family members.*

The information obtained in this way includes written records of names, dates of birth, addresses, history-based descriptions of phenotypes, information about medical conditions and biological inter-relationships among the family members (family pedigrees).

In this process information collected about the presence of genetic conditions in other members of a person's family enables conclusions to be reached about that person's genotype. These conclusions, of course, depend on our knowledge of patterns of inheritance and may be confined to statements of possibility. Nonetheless, the statement that a person has a fifty per cent chance of having a particular genetic condition because one of his parents had that condition amounts to genetic information. (Such statements, of course, were possible even before the existence of genes as such was known. At the time that Mendel developed his system, the existence of DNA was undiscovered. All that was known was that there were rules of heredity; the precise operation of the process was not to be discovered until the twentieth century.)

(b) *Direct observation of a person's phenotype.*

Such information would be a written record by the observer of the appearance and characteristics of the person. This may include the results of biochemical analyses of specific substances in the blood, urine, or other body fluids/tissues.

On the basis of this observation it may be possible to reach a conclusion concerning that person's genotype or the state of the person's chromosomes.

(c) *A laboratory-based gene products analysis.*

This type of analysis will determine the presence or absence of particular proteins. From this information conclusions can be drawn about the state of the genes which coded for those proteins.

For example the absence of the protein dystrophin indicates Duchenne's muscular dystrophy and hence indicates an abnormality in the gene coding for this protein. The written record of these proteins thus constitutes another type of genetic information.

24. A decision is therefore required as to whether principles pertaining to human genetic data should include all the above forms of genetic information, or whether they should be restricted to direct information about the DNA and the chromosomes. The arguments in favour of each position are:

(a) *In favour of including all forms of genetic information.*

Consistency requires that any regime be applied equally to all forms of genetic information, however it is obtained. There is no reason in principle why information about the DNA and the chromosomes, should be treated as more significant than other information which is effectively information about DNA, although obtained by a method other than DNA sequencing or DNA profiles.

Exclusion of family history information from the scope of any recommendations could mean that protection of a person from breach of confidentiality becomes dependent on the method by which the information was obtained. As a result an assessment of genetic risk derived from a family history could be used in a discriminatory way in the context of employment or insurance.

(b) *Against including all forms of genetic information.*

DNA testing is capable of disclosing a much greater range of information about an individual than indirect methods. Emerging chip technology will enable tests to be done for tens of thousands of sequences at a time, thus enabling information to be elicited relating to many conditions. DNA sequencing is therefore much more powerful and potentially informative than phenotypical observation or family history-taking.

The public perception also is that the results of DNA tests are matters of greater sensitivity than the selective, focused information obtained from, say, family history-taking. A person's genotype is seen as revealing something about his or her uniqueness as an individual. It therefore falls into a category of particularly personal information, needing special protection.

25. For the purposes of this Report, we recommend that the broader rather than the narrower definition of human genetic data be used i.e. the term 'human genetic data' will include all forms of genetic information, irrespective of whether they were obtained directly or indirectly. This is consistent with the position taken in the Report of the IBC on Confidentiality and Genetic Data.

IV. GENERAL ISSUES CONCERNING HUMAN GENETIC DATA

26. Human genetic data differs from general medical data about an individual in that they are of direct significance for the health of biological relatives, both living and unborn. Living relatives may, or may not, be aware that this information is being generated. Such family members may have a legitimate interest in the genetic material of their relative or in the information generated from testing. In addition, partners and spouses of family members may also have an interest because of concerns about the health of yet unborn children.

27. Exercise of the right not-to-know the results of genetic testing also needs consideration. Because families share genes, it can sometimes be very difficult to protect the right of one person to know about his or her genetic future while simultaneously protecting the right of a related person not to know.

28. The ownership of human genetic data requires further consideration. Are human genetic data a national resource or are they to be regarded as the property of the individual, group or community and a resource with economic potential?

V. PROCESSING AND STORAGE

29. The processing and storage of the human samples and genetic information derived from them can be summarized as follows.

- (a) The samples are labelled on collection and both the sample and the data derived are identified as belonging to a particular person. The sample may be destroyed or kept as agreed with the donor. Disposal should be by a method that is culturally appropriate.
- (b) The samples are not labelled on collection.
- (c) The samples are coded on collection and the identity of the person to whom the code relates is stored separately from the samples and derived data. Such samples are referred to as de-identified. Only specified persons have access to the identifying code. Samples are destroyed in a culturally appropriate manner or kept as agreed with the donor.
- (d) Samples are collected anonymously. There is no linkage between the donor and the genetic data.

Note that where human genetic data have been de-identified or anonymized, the opportunity for donors to receive the results of the testing has been lost.

Note also that sometimes even when the data have been de-identified or anonymized, the group identity of the person is retained along with the data. This situation needs also to be addressed as the group identity may be used for purposes of group discrimination/stigmatisation.

30. Access to genetic information in computerized databases requires special care to ensure that unauthorized persons do not gain access to it. This would include password protection and procedures for the protection of information during the upgrade and replacement of computers.

31. Access to genetic data after the death of the person from whom it was obtained presents special issues. Who, if anyone, should have access to these data? Such data may be of significance in the diagnosis and genetic counselling of children, grandchildren and other close relatives. A related issue which needs to be addressed concerns the collection of genetic data after the death of a person, for example from exhumed material.

32. Storage of and access to data obtained during prenatal screening also need clarification.

33. Where human genetic data are derived from direct analysis of the DNA, the obtaining of a sample of cells from the person concerned will be required. The ways in which blood, body fluid or tissue are obtained and the conditions in which they are stored or destroyed raise important ethical issues. Any statement of principles will therefore need to deal with the proper handling of the sample itself, as well as the handling of the human genetic data obtained from the sample.

34. Serious issues arise in relation to genetic research performed on archived collections of human samples, for example tissue samples which have been obtained at some time in the past for a different purpose. These historical collections can be important resources for research into human disease, as has been shown by the use in influenza research of samples of lung tissue taken from victims of the influenza epidemics of the early twentieth century. More recent collections raise issues of the rights for donors of tissue who may still be alive.

35. The IBC recommends that an instrument on genetic data should include provisions relating to the handling and storage of the human samples from which the data are obtained. This should include provisions dealing both with samples given for the purpose of genetic analysis as well as tissue originally obtained for other purposes.

VI. PURPOSES FOR COLLECTION

36. Genetic information is obtained for a variety of purposes which are not necessarily mutually exclusive. These may be classified as medical, social and research and development as follows:

(a) Medical

- *Diagnostic testing*
This refers to the identification of the cause of a disease.
- *Pre-symptomatic testing*
This refers to the identification of healthy individuals who may have inherited a gene for a late-onset disease, and if so will develop the disorder if they live long enough (e.g. Huntington disease).
- *Predictive/susceptibility testing*
This refers to the identification of healthy individuals who may have inherited a genetic predisposition that puts them at increased risk of developing a multifactorial disease, such as coronary heart disease or hypertension or monogenic diseases with incomplete penetrance such

as breast cancer due to mutations in BRCA1 or BRCA2 genes, but who, even so, may never develop the disease in question.

- *Carrier testing*
This refers to the identification of either healthy persons who may have inherited a mutated gene for a particular disease but which is not expressed in those persons or healthy persons who are carriers of balanced chromosomal rearrangements such as translocations and whose offspring are at risk of being affected.
- *Prenatal testing*
This refers to the genetic testing of developing foetuses which can be used to diagnose diseases or the likelihood an individual will develop a disease. It should be noted that this information will form part of the health record of the person to whom it pertains and that this raises issues as to the how, and at what age, the information will be communicated.

(b) Social

- Identification, for both forensic purposes and for the establishment of the relatedness of individuals, including paternity.
- Study of physical and psychological abilities.
- Identification of health risks for insurers and employers.

(c) Research and Development

- Determination of the sequence of the human genome;
- Population studies to establish, for example, genetic relations between different ethnic groups or the distribution of a particular gene;
- Determination of the genetic basis of a disease;
- Determination of genetic susceptibility/resistance to a disease;
- Localisation of disease genes on chromosomes
- Elucidation of the interaction of genes and environmental factors;
- Development of new drugs;
- Study of individual reaction to drugs.

37. The rationales for health-related testing and screening may be summarized as follows⁽²⁾:

(a) Screen and identify

- to treat,
- to counsel or educate,
- to isolate or segregate,
- to monitor or trace,
- to warn or protect third parties,
- to exclude, disqualify, transfer, discharge.

(b) Screen and not identify

- to count, survey, or track diseases,
- to study or research.

2. Jones D.G. Selected Legal Issues in Genetic Testing : Guidance from Human Rights. For the Health Canada Advisory Committee on Genetic Testing for Late Onset Diseases 2000.

VII PROVIDERS OF HUMAN GENETIC DATA

38. Persons providing samples for genetic testing may be grouped as follows (included also is an indication of the needs of each group).

- (a) Randomly selected individuals:
 - (i) Information provided for a single purpose,
 - (ii) Information provided for multiple use.
- (b) Individuals selected on basis of disease incidence / susceptibility / appearance.
- (c) Persons recruited as part of a group defined according to:
 - (i) Ethnic origins,
 - (ii) Geographic origins,
 - (iii) Behavioural attributes,
 - (iv) Chronological age.These groups would, in some countries, include persons taken into custody by the police.

VIII PRINCIPLES

39. The substantive principles which will inform the standards to be adopted with respect to the collection, processing, storage and use of human genetic data may be deduced from current human rights standards. These principles include:

(a) Respect for Human Dignity

This refers to the intrinsic worth and identity of human beings. It is included in the *Charter of the United Nations*, in the *Universal Declaration of Human Rights* and in Articles 1 and 2 of the *Universal Declaration on the Human Genome and Human Rights*. Genetic testing raises the concern that persons may be viewed as no more than their genetic characteristics. In addition Article 4 of the *Universal Declaration on the Human Genome and Human Rights* states that “the human genome in its natural state shall not give rise to financial gains”.

(b) Autonomy and Freedom

Genetic testing must not be undertaken without free and informed consent and wide-spread discussion for circumstances authorizing non-consensual genetic testing. The *Universal Declaration on the Human Genome and Human Rights* recognizes this in Article 5 and Article 9.

(c) Privacy and Confidentiality

This principle is recognized in Articles 7 and 9 of the *Universal Declaration on the Human Genome and Human Rights*. The principle is not an absolute one and the justification for the exceptions need to be widely-debated (see in this regard the Report of the IBC on Confidentiality and Genetic Data).

(d) Equality and Non-discrimination

Respect for human dignity means that individuals should not be burdened, mistreated or oppressed due to prejudicial attitudes about such attributes as biological status, race, religion, gender, age, disability. The application of this principle to genetic testing raises important questions concerning the grounds on which an individual is protected against genetic discrimination. It is recognized in Articles 2 and 6 of the *Universal Declaration on the Human Genome and Human Rights*.

(e) Justice and Solidarity

The multiple aspects of justice - 'distributive', 'procedural' and 'reparative' - have direct application to genetic testing. The concept of distributive justice is relevant to how society allocates the risks, benefits and burdens of genetic testing. These range from access to genetic testing services, to unduly burdening populations, to the allocation of genetic testing benefits and burdens between generations and between countries. It is particularly important that research efforts should promote health universally and so include developing countries. Article 17 of the *Universal Declaration on the Human Genome and Human Rights* draws attention to the need for States to respect and promote the practice of solidarity. The concept of reparative justice refers to the right of just reparations for those aggrieved or otherwise wronged by genetic testing initiatives. This is recognized in Article 8 of the Declaration. Procedural justice refers in part to fair process and procedure for dealing with alleged legal wrongs from genetic testing. It also raises questions about meaningful, inclusive and fair processes of decision-making on genetic testing at the individual, institutional and societal level.

40. Substantive principles alone are not sufficient to determine policy outcomes especially when there are conflicts between principles. An example of this would be conflicts between the principle of solidarity and the principle of confidentiality in relation to genetic information which may be of benefit to other family members. There may be cases in which breaching the right to confidentiality of an individual may appear to be justified in order to avert a serious risk to the life or health of another. Clear processes and fair procedures are essential for addressing, deliberating and mediating such conflicts. From the above discussion, a number of process values can be identified. These include:

- (a) Transparency of process and procedures,
- (b) Fairness of process and procedures,
- (c) Meaningful and inclusive involvement of society in decision-making,
- (d) Education of society,
- (e) Quality control (of laboratories undertaking genetic testing)

Article 13 of the *Universal Declaration on the Human Genome and Human Rights* indicates the responsibilities of researchers in carrying out their research. These include meticulousness, caution, intellectual honesty and integrity,

- (f) Monitoring and evaluation of processes

As stated in Article 13 it is important that public and private science policy-makers ensure that there are mechanisms for ensuring that proper processes are being followed.

IX APPLICATION OF THE PRINCIPLES

41. Acceptability of purpose for which human genetic data is collected

Guideline 1

(a) *Human genetic data may only be collected, processed and stored and processed for the following purposes:*

- *the provision of health care;*
- *research intended to further understanding of the structure and function of the human genome;*
- *the identification, in accordance with the provisions of national law, of those suspected of involvement in crime;*
- *the identification in the context of military service or for the purpose of identifying the victims of accidents or disasters, or for any other identification purpose authorized by national law and which is in conformity with the principles of international human rights law.*

(b) *Human genetic data may not be collected, stored or communicated for any purpose which is inconsistent with the principles set out in the Universal Declaration on the Human Genome and Human Rights.*

COMMENTARY

This initial principle stipulates that human genetic databases should only be established and operated for beneficial purposes. Thus, it would not be acceptable to collect or use genetic data for purposes of pursuing a programme of discrimination against a particular section of the population. Similarly, the collection of human genetic data in order to pursue a eugenic programme would be impermissible under this provision.

Research into behavioural genetics might fall under the health grounds stated above. If its focus were criminological or intended to measure and enhance intellectual ability or competence in the performance of tasks, then this might be considered to be research into the workings of the human genome. The acceptability of such work in any particular case would then depend on whether it was linked with an objective prohibited by the Universal Declaration on the Human Genome and Human Rights.

The provisions on identification raise issues of some complexity and sensitivity. Many countries now have forensic DNA databases, the aim of which is to link suspects with DNA samples obtained at the scene of a crime. The effectiveness of these databases as a means of apprehending offenders is well-established, but concerns have been expressed over the civil liberties implications of these measures. For this reason, a number of countries limit the circumstances in which samples for DNA testing may be taken or retained on forensic databases. The wording of the principle above recognizes that national laws will differ on this issue.

The collection of human genetic data about individuals for insurance or employment purposes is controversial and is the subject of legislative restriction in some countries. The principle stated above would preclude the testing of an individual specifically for these purposes, but would not preclude the communication of information about a genetic test which has been taken in a medical context. This issue will require further consideration.

The collection of human genetic data for the provision of health care to an unborn raises issues which also need addressing.

42. **Transparency of purpose**

Guideline 2

The purposes for which human genetic data are collected should be made clear to donors of data-producing samples by those responsible for the collection.

COMMENTARY

The aim of this principle is to prevent the building up of databases on the strength of false representations as to purpose or through the concealing of intentions.

43. **Role of education**

Guideline 3

Those promoting the collection of human genetic data should make every effort to inform the public of the purposes to which their collections may be put and to engage the public in debate over controversial issues. Health authorities, and those communicating with the public on issues of science and health, should endeavour to ensure an adequate level of public understanding of the benefits of the use of human genetic data in the context of health provision and health research before introducing new procedures.

COMMENTARY

A reasonable level of public understanding of human genetics is important if there is to be better understanding of the objectives of genetic research. This is particularly important in communities in which the public will be invited to participate in long-term research programmes into common illnesses. These research programmes, which could offer substantial benefits for our understanding of disease, require the participation of large numbers of people. An atmosphere of distrust of genetics could seriously affect these research projects. Specific mention is made of the responsibilities of communicators (journalists and others). The irresponsible reporting of science may threaten legitimate research; the responsibilities of journalists to report fairly and accurately need to be stressed.

44. **Public consultation and involvement**

Guideline 4

The establishment of any collection of human genetic data should be preceded by public consultation at the appropriate level. The views of interested organisations and individuals should be taken into account in the formulation of policies regarding administration and control of the database.

COMMENTARY

There is widespread acceptance of the need for public consultation in science policy, even if it is difficult to ensure that consultation is with the public in the wider sense rather than just with a narrow range of interested opinion. If science is to enjoy the confidence of the public and if abuses are to be prevented, science policy must be open to public scrutiny and take into account the views of those whose lives it will affect. The prolonged and major disputes over genetically-modified crops provides an example of how secretive or autocratic decisions as to what is best for people may be resented and may not accord with people's own vision of what is in their best interests. To avoid these misunderstandings, there should be consultation with a wide range of interests in civil society.

Guideline 5

All matters pertaining to the collection of human genetic data and any research which may be carried out on it should be subjected to independent ethical assessment of the same nature as is applied to any other biomedical research involving the use of human research subjects.

COMMENTARY

The provision relating to independent ethical assessment is an important one. Many countries now have research ethics committees, at national and local level, the function of which is to scrutinize research projects involving the use of human subjects. The collection of biological samples for DNA analysis is a form of research involving human subjects, and should therefore be subject to similar ethical controls. It should be emphasized that this scrutiny should not cease after the collection of data has been made. The subsequent use of the data and, also, of the samples themselves, needs to be subject to ethical scrutiny as does the storage of and access to both the samples and the data. Membership of ethics committees should be diverse, and include members of the community(ies) providing the samples.

45. **Cultural issues**

Guideline 6

- (a) *The human tissue from which genetic data are obtained, and genetic data itself, may be the subject of moral, social or religious beliefs. Those who collect human genetic data from communities holding such beliefs must give these convictions all due respect, endeavouring to do nothing to or with the genetic data, or the samples from which the data are derived, that would give offence to those from whom the samples and the data have been obtained.*
- (b) *National authorities promoting the collection of human genetic data from their populations should take into account the sensitivities of social, religious or ethnic groups within the population at large. Researchers must also observe the laws of the country in which their studies are conducted, as well as implementing the standards under which they work in their own country.*

COMMENTARY

This principle recognizes that there is considerable cultural sensitivity surrounding human tissue and genetic data. This matter has been substantially debated in the context of what has been called “gene prospecting”, the practice of collecting genetic data from isolated or indigenous populations. The genomes of such populations have the attraction to researchers of being relatively homogenous, which means that it may be possible to make a link between a particular genetic mutation and a phenotypical feature commonly observed within the community in question (such as immunity or particular susceptibility to an illness).

Community attitudes to these issues vary. Some governments have encouraged the collection of human genetic data in order to make this available to overseas researchers for financial gain. Consent is crucial here, and will be dealt with separately below, but the issue of respect for cultural and other sensitivities is important, and it is this issue which the principle seeks to address.

The Report of the IBC on Bioethics and Human Population Genetic Research⁽³⁾ (1995) had dealt with these issues. Particular attention has also been paid to this question by the ELSI Committee of the HUGO project, which has issued a statement setting out the responsibilities of those obtaining human genetic data from other communities.

46. **Counselling**

Guideline 7

- (a) *The genetic testing of an individual person for diagnostic purposes should be accompanied by suitable arrangements for the provision of relevant information both before and after a genetic test is undertaken and also for support in those cases where the test results may have serious implications for either the person tested or for others.*
- (b) *Consideration should also be given to methods of ensuring that those to whom genetic information is given are aware of the need to exercise caution in the passing on of this information to relatives who may themselves be affected by it.*

COMMENTARY

Testing a young person for a devastating monogenetic disorder would be irresponsible if no arrangements were made to explain the implications of such testing and to assist in dealing with the psychological consequences.

Parentage testing may also have serious consequences for individuals and for families, and it would be advisable to stipulate a counselling element for this form of testing.

47. **Validation and reliability**

Guideline 8

The obtaining of human genetic data from tissue samples should only be undertaken by persons whose expertise is established. Genetic tests should be properly validated for the purposes for which it is intended to use them.

COMMENTARY

It is important to ensure that if human genetic data is to be used for the purpose of diagnosis or identification it should be reliable and that claims for its diagnostic weight or its capacity to identify should be justified. This principle would have the effect of encouraging the setting out of laboratory standards. It would also discourage the use of tests supplied direct to the public (over-the-counter tests) unless such tests are approved by the appropriate authorities.

48. **Limitation of use**

Guideline 9

The genetic analysis of samples should be limited to the purposes for which information is legitimately acquired.

COMMENTARY

The aim of this principle is to prevent the analysis of samples beyond the scope of initially agreed purposes. If a person agrees to testing for one disease, it is not necessarily the case that he or she would consent to testing for another disease. Similarly, samples obtained for forensic identification purposes should not be subjected to testing aimed at diagnosing disease or determining behavioural characteristics.

3. See the *Proceedings of the Third Session of the IBC* (September 1995).

49. **Informed consent to diagnostic testing**

Guideline 10

- (a) *In the case of any diagnostic genetic test which may have serious implications for the welfare, psychological health or family relations of the person tested, no test shall be administered unless the person consents after being informed that the test is a genetic one and is informed of its implications. Consent may be given for the future testing of samples should new tests be indicated or become available.*
- (b) *Where a person to be tested is unable to give a valid consent on the grounds of minority or on the grounds of mental incapacity, then the consent should be obtained of any person legally authorized to give such consent on behalf of the minor or incapable person after full account has been taken of the present and future impact which the test might have upon the tested person.*

COMMENTARY

Genetic testing is likely to become increasingly common as the genetic component of disease is further identified and as progress is made in pharmacogenetics. Many tests which reveal genetic information will not have a great deal of significance for the person tested (a blood group test, for example, reveals genetic information in that blood grouping is based on genetic factors. Other tests, however, will have major implications, both for the individual and for relatives. The principle stated above sets out the consent requirements. For practical reasons, it would be unrealistic and unnecessary to require that there be specific consent to the genetic component in any test unless the consequences of this are sufficiently serious to justify this.

50. **Free and informed consent in research**

Guideline 11

- (a) *A research sample may be collected from a person only after the person has first been given a sufficient explanation in an appropriate style and language of:*
- *the purpose for which the sample is collected;*
 - *the source of funding for the research;*
 - *the type of use which will be made of the sample or any information derived from it; and*
 - *any other implications which the collection and subsequent use of the sample might reasonably be expected to have for the person providing it.*
- (b) *The consent should be expressed in writing unless there are cultural reasons for not doing so, in which case other possibilities of publicly recording consent should be explored.*
- (c) *Donors of samples for research purposes should not be subjected to any pressure or improper inducement in order to secure their consent. A person who refuses consent should not suffer any adverse consequences as a result of this refusal, and all potential donors should be given an assurance to this effect.*
- (d) *Samples donated for research purposes should not be made available for other purposes, such as police use, and national legislation should seek to prevent such use.*

COMMENTARY

The consent requirements in the research context are more rigorous. A difficult question in this area is that of whether fresh consent has to be obtained if new research of a different nature is to be conducted on samples originally given for another form of research. A system which required fresh consent would be extremely cumbersome and could seriously inhibit research and it is for this reason that a system of “blanket consent” covering all forms of future medical research might be preferable, provided that the consent given in the first instance explicitly recognizes this. This is envisaged in the wording above which requires that information be given as to the “type of research” involved. It would, of course, be ethically impermissible to carry out non-medical research on samples donated solely for medical purposes.

51. **Obtaining research samples from those unable to consent**

Guideline 12

Where consent to the collection and use of a research sample is sought from a person who is unable to give a valid consent on the grounds of minority or on the grounds of mental incapacity, then the consent should be obtained of any person legally authorized to give such consent on behalf of the minor or incapable person after full account has been taken of the present and future impact which the test might have upon the tested person. Such consent should only be given if any adverse implications for the tested person are negligible and if this conduct of research in these circumstances is permitted by national law.

COMMENTARY

There has been considerable discussion in biomedical ethics of the legitimacy of conducting research on minors and people affected by a mental incapacity. While there continues to be some disagreement on the matter, a defensible position is to allow such research to be carried out provided that (a) there is no other way of conducting the research; (b) the research is of clear benefit to the community and, in particular, of benefit to the category of persons from which the subject is drawn (that is, the research is of benefit to minors or to those affected by any condition from which the mentally incapable person suffers); and (c) there is negligible risk or discomfort for the participant.

52. **Withdrawal of informed consent**

Guideline 13

Providing samples and derived data have been stored in an identifiable manner, donors may request that those samples and any human genetic data derived from them be returned to them or destroyed.

COMMENTARY

This embodies the principle endorsed in the major codes relating to the use of human subjects in research that the subject may withdraw at any time during the course of the research. A possible objection to this is the inconsistency which this involves with the principle of donation. An alternative approach would be to treat the giving of a sample as amounting to a donation, thus transferring full ownership and control over the sample to the researcher.

In favour of recognizing a continued interest on the part of the provider of the sample is the proposition that people do not wish to relinquish control over genetic information about themselves. If the researcher has complete and ultimate control over the sample, then it might be possible to extract from the sample information which the “donor” might wish to keep confidential.

53. **Confidentiality/Anonymity**

Guideline 14

Donors of samples for genetic testing, both individuals and communities, should be informed prior to giving consent whether their samples and genetic data will be identified, coded but identifiable, or not identifiable (anonymized), and the meaning of these terms should be clearly explained.

COMMENTARY

This recognizes the various forms in which samples might be stored and requires that this be explained to the donor of a sample. Issues concerning a population identifier need further discussion.

54. **Fate of material**

Guideline 15

Donors of identified samples must consent to whether their samples, extracted DNA, and genetic information will be stored or disposed of. If human samples are destroyed it must be done in a way that takes into account any sensitivities which donors may have regarding this process. If stored, the donor must be told where the sample will be stored. Tissue or DNA should not be exported without the consent of the donor.

COMMENTARY

The aim of this principle is to prevent subsequent use of samples to which the donor might reasonably have objected. In particular, insensitive destruction of tissue is a matter on which people may have strong views, particularly if the tissue is obtained through post-mortem examination. The exporting of samples is also a matter on which people may be sensitive.

55. **Sharing information**

Guideline 16

If those conducting research do not envisage informing the donors of samples of any results obtained from testing individual samples, then this fact should be communicated to the donor prior to obtaining consent for the taking of the sample. Where the overall results of research projects can be communicated to the donors, then it is good practice to arrange for this to be done, either individually or through the use of appropriate means of mass communications both to individuals and to families and populations.

COMMENTARY

The issue of “feedback” to participants in research is an extremely difficult one. In small research projects, involving relatively few participants, it is desirable that there should be communication to individual participants of any information which emerges which is relevant to their health, provided, of course, they have indicated that they would wish to hear it or, in the particular circumstances, they might reasonably be expected to wish to have this information. This practice, however, is not followed in large-scale epidemiological research projects, as this converts the nature of the exercise from a research one into a mass screening undertaking. The latter might have quite different budgetary and counselling implications and would have the effect of preventing some forms of research from being done in the first place. For this reason, the duty to inform is restricted in this principle.

56. **Post-mortem samples**

Guideline 17

The taking of samples from the dead for the purposes of obtaining human genetic data should only be undertaken in the following circumstances:

- (a) *the sample is required for the purposes of criminal or civil justice and has been legally authorized for these purposes; or*
- (b) *the taking of the sample has been authorized by the next-of-kin of the deceased person and there is no evidence that the deceased person might be expected to have objected to the purpose for which the sample is sought; or*
- (c) *the person from whom the sample is taken has been dead for such a length of time that there is no reasonable prospect of offence being given to identifiable descendants. Where there is a contemporary community which reasonably considers itself to be linked with human remains in this category, then the consent of such community should be obtained before a sample is taken.*

COMMENTARY

It is generally accepted that the dead should be treated with respect, the content of that respect varying from culture to culture. The DNA testing of the dead is potentially an infringement of privacy rights which the deceased enjoyed during his or her lifetime. There are, however, legitimate purposes which might be served by testing the dead (these may be research purposes or they might be purposes connected with the diagnosis of illness in a person related to the deceased). In these circumstances, unless it is known that the deceased held an objection to the procedure, there might be a presumption of altruistic intent and testing might be permissible.

The provisions relating to those who died a considerable time ago recognize that the moral claims which the dead may have are weakened by the passage of the years. At the same time, there are some communities which feel strongly about even very old remains and this feeling should be respected.

57. **Historical or archived collections**

Guideline 18

The obtaining of human genetic data for medical research purposes from historical collections of human tissue should only be undertaken without the consent of the donor of the sample if the sample is anonymized. If the sample is not anonymized, the consent of the donor should be obtained, provided that the donor can be traced with reasonable effort.

COMMENTARY

Historical collections of tissue are of great importance in medical research. The denial of these collections to researchers would impede medical progress and for this reason their accessibility to researchers should not be barred. If it is possible to identify the donor and trace him or her without excessive difficulty, then that would be consistent with an approach which recognizes the importance of consent in general. However, in many cases this will not be possible; in which case the samples should be anonymized before being used without consent.

58. **Rights of children**

Guideline 19

When children are the providers of samples for DNA analysis, investigators must ensure that:

- (a) the children and their parents, guardians or caregivers have been fully informed, and that the information for children is presented in a style appropriate to their age;*
- (b) where a child has the competence to understand the nature, risks and consequences of the proposed procedures the consent of the child is obtained;*
- (c) where a child lacks the necessary competence:
 - (i) the child's parents or legal guardian gives permission for the child's participation;*
 - (ii) the child's assent must be obtained;*
 - (iii) the child's refusal is respected.*
 - (iv) presymptomatic testing of children at risk of late onset diseases such as Huntington's disease is not permitted even when the parents or legal guardian consent to or request this.**

COMMENTARY

The special vulnerability of children requires that special ethical considerations should be in place for reviewing children's involvement as providers of genetic data. The recognition that children are persons in their own right with their own unique set of interests requires that particular attention must be paid to the provision of information to children, the gaining of their consent and their right to refuse participation.

59. **Collection of samples in other countries**

Guideline 20

The collection of samples by researchers in a country other than their own, particularly where the researcher(s) are from a developed country and the collection is taking place in a developing country, must only be undertaken after appropriate consultation with the people of that country and according to the laws of both that country and the country of the researcher. Where ethics committees exist in the country where collection is to take place, permission must be sought from the relevant committee as well as an ethics committee in the country of the researcher. The country of origin of the samples should be disclosed in all publications resulting from the research.

COMMENTARY

Persons in developing countries are particularly vulnerable to exploitation in the collection of biological samples for genetic testing. The research community must take particular care to ensure that the rights of individuals and communities providing samples are protected. Issues concerning ownership of the information and financial rewards from the use of the information must be considered. In addition, Article 19 of the *Universal Declaration on Human Rights and the Human Genome* makes it clear that developing countries must benefit from the achievements of scientific and technological research.

60. **Collection of samples from indigenous peoples**

Guideline 21

The collection of samples from indigenous peoples must only be undertaken after appropriate consultation with those peoples according to their customs and protocols and according to both the laws of their own country and the laws of the country of the researcher.

COMMENTARY

Indigenous peoples are particularly vulnerable to exploitation in the collection of biological samples for genetic testing. The research community must take particular care to ensure that the rights of individuals, families and communities providing samples are protected. Issues concerning ownership of the information and financial rewards from the use of the information must be considered.



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