FUNDAMENTAL PRINCIPLES
OF RESEARCH ON THE HUMAN GENOME

JUNE 14 , 2000

Council for Science and Technology, Bioethics Committee

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Basic Ideas

1. **Science**, as one of the intellectual activities of humanity that aims at the pursuit of truth, underlies the future development of human society. Freedom of research, which is the pillar of science, is a part of freedom of thought which, in turn, is a core component of fundamental human rights. Science, however, is not an autonomous entity which is independent of society but one which is confined within the sphere of human society. Thus, scientific research should conform to the respect for human dignity, should interact with various social factors, and should fully understand that there might be oppsitions or conflicts between scientific interests and other social intersts.

2. **Life science** aims at the elucidation of phenomena concerned with the life of living organisms, especially at understanding humanity from a biological point of view. The applications of the results of research in life science, including medical treatments and agriculture, have made a significant contribution to the progress of the health and welfare of humanity. The application of findings derived from research on the human genome, in particular, shed light on the possibility of the prevention and cure of diseases which have defied traditional medical approaches. It is a concern that progress in life science, on the other hand, may precipitate various problems at the interface with society. We, the humanity, have the history in which we have seriously neglected considerations of human dignity and human rights in the name of research. After the Second World War, these acts of violation were reflected on with deep regret. So that such blunders of the past are not repeated, all nations and related international organisations have been constantly makin various efforts from the viewpoint of bioethics, beginning with the pronouncement of the Nuremberg Code. Medical doctors and researchers, too, awakened by the Declaration of Helsinki and other instruments , have come to realise that human dignity and human rights should be respected in their research. In relation to research on the human genome which is the focus of the present Fundamental Principles, “the Universal Declaration on the Human Genome and Human Rights” was adopted in 1997 by the General Conference of UNESCO (the United Nations Educational, Scientific and Cultural Organisation). This Declaration was endorsed by the General Assembly of the United Nations and accepted in various countries as the first instrument of universal ethical principles concerning research on the human genome.

3. **Research on the human genome** made rapid progress due to the “Human Genome Project”, which was effectively started in earnest in 1990. The research on the human genome aims to analyse the structures and functions of the human genome, investigate the biological functions of humans, and understand the mechanisms of human life. Based on this information, the human genome research aims to contribute to the protection of human life and health and to the

prevention and cure of diseases. Great strides have been made in research on genetic polymorphism, in particular, due to the accelerated decoding of DNA sequences. Research on the genetic information collected from individuals enables the aetiology of diseases to be determined, novel strategies for the prevention, diagnosis, and cure of diseases to be developed, and also for new medicines to be developed. Some of the findings generated by this research are now being put to practical use.

4. **Research on the human genome and its outcomes**, however, may elicit serious ethical, legal and social issues because, on the one hand, they may lead to the manipulation of 'life' and, on the other hand, they may bring about the risk of violating the dignity and human rights of an individual on account of his/her personal genetic characteristics. Thus, it is imperative that all these issues are guarded against and that the full understanding of the public is obtained in regard to research activities on the human genome and the application of the outcomes of the said activities.

5. **"The Fundamental Principles"** were established in order that research on the human genome is conducted in an appropriate manner without violating human dignity or human rights. The principles are mainly oriented to researchers and medical doctors concerned with the human genome. These principles also provide the basis for the understanding of the research on the human genome which the public, especially the persons providing research samples, their blood relatives and families, are expected to have. It should be noted that research on the human genome is intricately related to the diagnosis and treatment of diseases based on the genetic information it provides. National guidelines on genetic diagnosis and gene therapy need to be established separately in addition to the "Fundamental Principles of Research on the Human Genome".

Chapter 1 The human genome and the research on the human genome

Principle 1 The significance of the human genome

1. The human genome is the heritage of humanity.
2. The human genome is the blueprint of human life. It provides all members of humanity with a biological basis as a human being and it also underlies the uniqueness and diversity of each human being.
3. The existence of a human being is not determined only by his/her genome.
4. Conveying the fundamental information of human life from parents to children, from children to grandchildren, and so on, the human genome builds the fundamental structures and functions of an individual as a human being. At the same time, however, the human genome is acted on by various influences in the natural and social environment.

Principle 2 Diversity of the human genome and the dignity and human rights of individuals

The human genome differs from one individual to another. The diversity of genetic characteristics signifies the originality and uniqueness of each individual and the plurality of humanity as a whole. Thus, it is imperative that, regardless of their genetic characteristics, all individuals or groups of individuals are respected for their dignity and human rights, and that they are equal to one another and are not subjected to any form of discrimination.

Principle 3 Due consideration to ethical, legal, and social issues

It is considered that since research on the human genome and the applications of its outcomes have the potential to significantly change the way in which human beings think about human life and living, it may have an extremely serious impact on society. Therefore, due consideration should be given to ethical, legal and social issues in research and its applications.

Principle 4 Participants, their families and blood relatives

Research on the human genome inevitably requires research samples that are provided by human subjects. Therefore, this type of research can be conducted only with a high regard for the dignity and human rights of the persons providing research samples (hereafter, “participants”), their families and blood relatives.

Chapter 2 Rights of participants

Section 1 Informed consent

Principle 5 Basic Conditions

1. A research sample may be collected from an individual subject for research on the human genome only after having given the participant sufficient explanation beforehand and having obtained the consent of the participant on his/her own free will (informed consent).
2. The consent should be expressed in writing.
3. Individuals who are requested to provide a research sample but do not consent to that request should not be disadvantaged for his/her refusal.

Principle 6 Individuals who have not the capacity to consent

In the event that research on the human genome is to be conducted with participants including individuals who have not the capacity to consent, it should be shown in advance that the research in question requires a research sample to be obtained from the said individual, and his/her informed consent should be obtained from his/her representative.

Principle 7 Diversity of research

1. In the procedure of obtaining informed consent, the information and explanation about the research should be given using the most appropriate method for the objectives and details of the research, in view of the fact that research on the human genome, especially identification of genetic information of individuals, is associated with a high risk of ethical, legal and social issues.
2. In all research projects, the procedures used to obtain informed consent should be clearly described in the research plan and undergo prior review by the Ethics Committee.

Principle 8 Comprehensive consent, and unlinked and unidentified research samples

1.
    (a) If a participant consents to provide a research sample for a genome analysis in a particular research project and, at the same time, anticipates and consents to the use of the same sample in other genome analyses or related medical research, then the research sample may be used for the latter “studies aimed at other purposes”.

    (b) In this case, sufficient information, which clearly outlines the anticipated objectives of the research at that point in time, should be given to the participant so that s/he can thoroughly understand the significance and consequences of the fact that the sample provided will be used in “studies aimed at other purposes”, as defined in the preceding sub-paragraph.

    (c) In the cases described in the preceding two sub-paragraphs, protocols pertaining to the management and protection, including anonymity, of genetic and other personal information of the participant should be explained in detail, and strict confidentiality should be guaranteed.
2. If it is expected that the sample provided will be handled anonymously, the method of explanation used in the procedure of obtaining informed consent may be appropriately simplified on the condition that it is guaranteed that the sample can not be linked to the subject.
3. In the cases described in the preceding two paragraphs, the comprehensive consent and the simplified method of explanation can be used only when the source of the research sample to be used, and the actual protocols and method used in the procedure of obtaining informed consent are described in the research plan and when these issues are reviewed in advance by the Ethics Committee concerned.

Principle 9 Existing samples

1. Existing samples, which have already been provided prior to the entry into force of the present Fundamental Principles and for which informed consent was not obtained at the time of

provision, may be used only after consent is newly obtained.

2. Existing samples, which have already been provided prior to the entry into force of the present Fundamental Principles and for which informed consent was obtained at the time of provision, may be used only within the framework of the said consent.

3. Notwithstanding the preceding two paragraphs, in case that research is required to be undertaken using an existing sample for which informed consent was not obtained at the time of provision or in case research which is beyond the framework of the obtained consent is required to be undertaken, the said sample should not be used prior to a review by the Ethics Committee. The Ethics Committee should determine the conditions for the use of existing samples, including the necessity of newly obtaining informed consent, by taking the following points into consideration: the anonymity of the sample, the possibility of linking the sample to its participant, the characteristics of the sample, the research plan and details of the said research, potential impact on the participant, etc., measures for the protection of personal information, and so on.

4. In the case of existing samples, personal information, including genetic information, should be kept strictly confidential and rigourously protected. Researchers and research institutions should establish a system for the control of personal information.

5. Existing samples, which are entrusted to organisations specialised for the storage, such as human material banks, or which are already on the market, may be dealt with in the same manner as for the general samples for scientific research.

Principle 10 Withdrawal of informed consent

1. The consent to donate a sample for research may be withdrawn providing that the said sample can be linked to its participant.

2. A participant should not be disadvantaged even if s/he withdraws his/her informed consent.

Section 2 Genetic information of participants

Principle 11 The protection and control of genetic information and establishment of a control system

1. The genetic information of participants should be kept in strict confidence and under full protection.

2. Researchers and research institutions should keep in strict confidence and control the personal information of the participants, identifying the information with which the participant can be linked to the research sample, and the genetic information of the individuals that is obtained as a result of the research. They should also protect the said information with the utmost caution. To this end, research institutions should establish and consolidate systems

and procedures required to control and protect genetic information along with other personal information and identifying information.

3. All research institutions should make the importance of protecting the personal information in research on the human genome common knowledge to all researchers and personnel involved in the research. The significance of personal information and the necessity for its protection should be thoroughly understood by all.

Principle 12 Leakage of personal information

1. Research institutions and researchers should take necessary measures in order to prevent the leakage of personal information.
2. In the event of the leakage of personal information, firm disciplinary measures, including demotion, should be taken against the person(s) who leaked the information, the researchers undertaking the said research, the custodians of the personal information, the director of the research institution and other personnel associated with the leaked information.
3. Any individual whose personal information is leaked or who sustained damage from the said leakage is legally entitled to receive compensation or indemnity.

Principle 13 Right to be informed

A participant has the right to be informed of his/her genetic information resulting from the research.

Principle 14 Right not to be informed

A participant has the right not to be informed of his/her genetic information resulting from the research. The findings of the research may not be made known to the participant against his/her will.

Principle 15 Disclosure of information to blood relatives

1. In principle, blood relatives or families of participants may be informed of the genetic information of the participant only when a participant gives his/her own permission. Personal information pertaining to a participant may not be disclosed to his/her blood relatives or family against his/her will.
2. Notwithstanding the principle described in the last paragraph, in case that the genetic information obtained by research may lead to an interpretation that a portion of the genetic characteristics of the participant is or, is supposed to be, connected to the aetiology of a disease, this interpretation may be disclosed to his/her blood relatives following authorisationby the Ethics Committee only if a preventive measure or a cure has already been established for the disease in question.

Principle 16 Prohibition of discrimination

The genetic information of a participant forms the basis of his/her diversity as a member of humanity. The participant should not be subjected to any discrimination on account of any genetic characteristic in his/her genetic information that is obtained from the research.

Section 3 Other rights and interests

Principle 17 Gratuitousness and related principles

1. All research samples should be provided gratuitously.
2. In the event that an outcome obtained as a consequence of a research project becomes the subject of intellectual property rights or other rights, these property rights are not attributed to the participant.

Principle 18 Reparation for damages

A participant has the right to receive compensation or indemnity if s/he incurs damages in the process of research on the human genome or in relation to that research.

Principle 19 Social and psychological support

On providing a research sample, or on knowing or being informed of the results of the research, a participant, his/her blood relatives and family have access to all pertinent social and psychological support, including genetic counselling.

Chapter 3 Fundamental requisites of research on the human genome

Principle 20 Human dignity and freedom of research

1. Research, which is contrary to human dignity, shall not be undertaken.
2. Freedom of scientific research should be respected.
3. Research on the human genome and its applications should be conducted with full respect for human dignity and human rights.

Principle 21 Requisites of research and establishment of research plans

1. Research on the human genome should be designed so that useful and beneficial coutcomes are estimated in the fields of biology, genetics and medicine.
2. Research on the human genome should be conducted on the basis of a clear and detailed plan of research.
3. Information regarding DNA sequence data should be released into the public domain.

Principle 22 Establishment and observance of protocols for conducting research

Research on the human genome is diverse depending on the objectives and subjects of each project. Adequate research protocols regarding the specific conditions detailed in the research plan should be established while respecting the present Fundamental Principles of Research on the Human Genome. All researchers and personnel involved in the implementation of the research should observe the said protocol.

Principle 23 Ethics Committee

1. On undertaking any research on the human genome, its research plan should undergo a prior review by an independent, multidisciplinary and pluralist Ethics Committee.
2. The Ethics Committee should examine a submitted research plan on the human genome mainly from the ethical, legal and social points of view in addition to its scientific merit and, comprehensively evaluate whether the implementation of the project should be approved or not.
3. The Ethics Committee should guarantee its transparency in its organisation and reviewing deliberations.

Chapter 4 Relationship with society

Principle 24 Understanding and support by society and accountability

1. Research on the human genome makes a significant contribution to the life and health of humanity and each individual, and to the welfare of society.
2. Public support of the advancement of research on the human genome with recognition of the role played by the said research in society now and in the future is desirable. This support should be underpinned by a sound understanding of the present Fundamental Principles of Research on the Human Genome (especially the significance of the human genome outlined in Principles 1 to 3).
3. All people involved in research on the human genome have a general accountability to society on all aspects of the research in order to improve the understanding and recognition of the public as described above.

Principle 25 Public disclosure of outcomes of the research and return of benefits to society

1. All beneficial outcomes in the fields of biology, genetics and medicine obtained from research on the human genome should be returned to society. In principle, they should be disclosed to the public.
2. All outcomes of research on the human genome should be employed for the development of science, the elimination of human suffering, the prevention and cure of diseases, and the

improvement of health.

Principle 26 Appropriate measures

Appropriate measures should be taken so that research on the human genome can be adequately and effectively promoted in accordance with the "Fundamental Principles of Research on the Human Genome". In addition, general, adequate and prompt decisions and countermeasures should be taken regarding the various ethical, legal and social issues that may arise from research on the human genome and its outcomes.

Principle 27 Dissemination of education and provision of information

Bearing in mind that research on the human genome will have a serious impact on life, living and the future of humanity and each individual, education on the human genome and its research as well as bioethics should be widely promoted. Endeavours should be made to spread information concerning research on the human genome and its applications.

Supplementary provision

These "Fundamental Principles of Research on the Human Genome" should be revised as appropriate in the light of the actual progress of research on the human genome and the understanding of such research by the public and social trends in this regard.