REPORT
on the collection and use of genetic data

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The aim of the present report is to prepare the ground for a debate on the principles that should govern the collection and use of genetic data. The term genetic data refers to any information collected or resulting from genetic tests or analysis of the genetic material of one or more persons. We use the term genetic test when the aim is to collect medically relevant information and the term genetic analysis when the aim is to verify a person's identity or parenthood. Regardless of the aim they are collected for, genetic data characterizing specific persons or groups of persons constitute "sensitive" personal data as they may lead to social discrimination and group stigmatization.

The main ethical questions arising with regard to the collection and use of genetic data are: the extent of information to be provided to and the procedures to ensure the consent of the examined person or group; how to secure the confidentiality of genetic information and what is the desired level of confidentiality; how to safeguard freedom of research; whether genes can be patented (e.g. the genes predisposing to a specific disease) and, if yes, who is the rightholder (the scientist or the subject of research). The answers to these questions are not universal but depend directly on the type of genetic data (e.g. identifiable or anonymous), their possible use and the socio-cultural context of genetic testing or analysis. The present report attempts to introduce the questions regarding the collection and use of genetic data.

The Commission has already addressed part of the issue in its report on genetic data collected in criminal investigations which, in fact, covers most of the genetic analysis spectrum. Thus, in the present report we decided to focus our interest on the collection and use of genetic data for medical purposes (genetic tests).

In order to facilitate the debate on genetic data, the first part of the report includes: a) a short introduction to the basic principles of genetics, b) the main differences and similarities between genetic and medical data, c) a classification of genetic disease and tests, and, d) a brief outline of genetic databases. The second part discusses the

main ethical problems arising with respect to genetic tests and provides a summary of potential answers. The third part discusses the legal approach to genetic tests from the viewpoint of a) international and foreign law, and, b) domestic law. Finally, in the fourth part, we attempt to present an ethical framework which we consider appropriate for the collection and use of genetic data based on the previous parts.

PART ONE

A. BASIC PRINCIPLES OF GENETICS

Under normal circumstances, all human somatic cells contain 23 pairs of chromosomes in their nuclei. Chromosomes are the structural vehicles of DNA that carry the genetic information that is "written" on it and participate in two basic functions: the transmission of genetic information from cell to cell (cell division) and from one generation to the next (production of gametes).

Of the total 46 chromosomes that constitute the normal human karyotype, 23 chromosomes (one of each pair) originate from the maternal ovum and 23 originate from the paternal sperm. Thus, the gametes contain half of the genetic information present in each somatic cell. When the nucleus of the ovum merges with the nucleus of the sperm, the number of chromosomes doubles and, thus, the zygote and the resulting organism acquire the normal karyotype of the species.

Chromosomes are divided in autosomes (22 pairs) and sex chromosomes (23^{rd} pair). Usually, the normal human karyotype is reported as 46XX or 46XY depending on whether it refers to a female or male respectively. That is, the total number of chromosomes and the constitution of the 23^{rd} pair which is either XX or XY. Chromosome Y occurs only in males. As mentioned above, during gamete production, the number of chromosomes is halved, meaning that the chromosomal constitution of spermatozoa with respect to sex chromosomes may be either X or Y while the ovum always carries an X chromosome. If a spermatozoon carrying a Y chromosome fertilizes an ovum (X), the descendant will be male (XY) while if the spermatozoon carries an X chromosome, the descendant will be female (XX). In males, therefore, chromosome X always originates from the mother while females inherit one X chromosome from each parent.

Hundreds or thousands of genes are arranged end-to-end along chromosomes on the DNA. Each gene is located on a specific chromosome and its location is unique. DNA consists of a sequence of four bases (A=adenine, T=thymine, C=cytosine, G=guanine). The arrangement of these bases in triplets represents the genetic code. Some triplets correspond to an instruction to recognize certain amino acids while others signal the "start" or "end" point of message transcription[1].

All individuals in a given species have the same set of genes but the precise DNA sequence of given genes varies between individuals. This variation in individual genes may correspond to the substitution of one base by another (i.e. A→T) or the deletion or insertion of a base, etc. Sequence variations in DNA can have varied effects. We use the term sequence polymorphism when the specific variations do not affect gene function and the term mutations when they do. Some mutations may lead to disease; others predispose or protect or modify the severity of a disease.

Finally, as far as the distribution of genes to various chromosomes is concerned, chromosome Y carries very few genes whose function affects mainly the process of spermatogenesis, that is male fertility. In contrast, chromosome X is the richest chromosome in genes since it carries about 5% of the total genetic information of the human organism. Given that the human karyotype for females and males is XX and XY respectively, women carry the richest chromosome in genes twice as compared to men. Remarkably, however, "sex equality" is ensured through a mechanism of random inactivation of one of the two chromosomes X present in each female somatic cell. As a result, both sexes carry only one active chromosome X in each cell. Since males inherit their single X chromosome from the mother, they always express the maternally inherited genes. Females, instead, express a mosaic of maternal and paternal genes in chromosome X.

B. GENETIC AND MEDICAL DATA

Research on the genetic basis of disease aims mainly at the development of specialized genetic tests to facilitate the diagnosis, prevention and treatment of medical conditions in the hope of reducing mortality and morbidity rates and improving quality of life. The benefits expected from genetic research and the

[1] The terms "mapping" and "decoding" of genome refer to the effort to record the unique location of each gene and the base sequence of the DNA respectively in order to read all the transcriptions.

integration of its techniques in clinical practice are no different than those expected from medical research in general.

However, genetic testing may help to investigate the extent to which a person's current medical condition is influenced by specific gene mutations or to detect a genetic predisposition to a specific disease that will develop in the future and, in case of genetic disease-carriers, to evaluate the probability of affected descendants. It is argued that if such "sensitive personal information" is left unprotected, there may be adverse consequences in health care, employment and insurance. Moreover, the knowledge and disclosure of such information may significantly limit personal reproductive choice and/or lead to social discrimination and stigmatization.

Although the consequences of the ever increasing amount of genetic information cannot be foreseen with any degree of accuracy, the scientific community and several social agencies have expressed concerns over the principles that should govern the use of this information as well as the circumstances in which the collection of such information is indicated.

Given that the use of medical data is currently subject to regulation, a critical question arose as to whether the data collected and/or resulting from genetic tests, i.e. genetic data, constitute even more sensitive personal information as compared to medical information and whether, as a result, they call for special protection.

To answer this question, it should first be clarified whether genetic data are significantly different from medical data and whether the personal and sensitive nature of this information is susceptible to greater abuse.

The term genetic data normally refers to information collected or resulting from genetic analyses carried out on individual DNA samples. However, information about a person's genetic makeup can also be obtained by biochemical tests designed to detect the production of enzymes or proteins revealing the occurrence of particular genetic mutations in this person's DNA. Moreover, a family's medical history can lead to conclusions about an individual's genetic makeup. For example, if a male child suffers from hemophilia A[2], it may safely be concluded that: a) the disease was inherited from the mother who is thus a carrier of the disease-causing gene, and, b) there is a 50% probability that the mother's sisters, if any, are also carriers themselves.

[2] This disease is sex-linked, see pp. 7-8.

Yet, although in some cases genetic information does not differ significantly from medical information, the discussion about genetic data does have some unique implications. Firstly, genetic information about a person always reveals, even if partially, information about third parties (blood relatives) and thus it has a more direct impact on the family of the examined person while access by third parties (employers, insurance companies) to such data brings forth the question of privacy not only of the person involved but also of persons genetically linked to him/her. Secondly, much of genetic information is probabilistic since genetic predisposition to a disease does not necessarily mean that the person in question will definitely develop it and, even if they do, it is impossible to predict the exact moment the disease will be manifested. Furthermore, even when a severe genetic disease is highly probable, this does not cancel either the probabilistic nature of the information or the possibility of mistake. And yet, these inherent risks of genetic information are often ignored and genetic predictions are often mistakenly seen as deterministic. The overestimation of the importance of genetic information is mainly due to confusing the precision of information at the level of DNA (accuracy of data) with the precision of predicting a disease. This confusion may lead to restriction of individual reproductive choice - and, generally, freedom - or to social stigmatization and discrimination. Apart from confusion, however, there are also the proponents of the doctrine of genetic determinism who fail to acknowledge the influence of the natural, social and economic environment on character in relation to behavior or health. Ignoring the impact of environmental factors on the manifestation of such traits has at least two implications: a) it shuns personal responsibility since people cannot be held responsible for their actions for they do not choose their genetic makeup which determines them, and, b) it precludes measures of social solidarity and equity since no intervention in the socio-economic environment can change or improve the manifestation of such traits. Such theories are not only scientifically unsound but entail objectionable social practices and attitudes and, therefore, lead many people to argue that genetic data call for specific protection regimes as compared to medical data.

C. GENETIC DISORDERS and GENETIC TESTS

The term genetic disorder refers to abnormalities due to gene mutations or deviations from the normal karyotype. In these cases, the mutation or the chromosomal abnormality is present in the zygote and thus can be detected in any cell of the organism at later developmental stages and, naturally, may be transmitted to offspring via the gametes.

Some genetic disorders are not hereditary. This happens when gene mutations or chromosomal abnormalities occur after conception in a somatic cell. In these cases, genetic disorders are not transmitted to offspring, are not detected in parents and, generally, have a limited distribution in the organism[3].

Genetic disorders that are due to chromosomal abnormalities may occur as deletions or excesses of entire chromosomes or chromosomal segments in the cells of the organism. Given that there are hundreds of genes on each chromosome, such disorders have more or less severe clinical symptoms. The existence of chromosomal disorders in the zygotic cell often prevents the implantation of the foetus. Even when the foetus is implanted, embryonic development is usually abnormal and leads to embryo loss. However, embryos with certain chromosomal abnormalities do survive, the classic example being the Down Syndrome which is due to an extra chromosome 21 (trisomy 21).

Monogenic disorders are hereditary diseases due to single gene mutations. The majority of monogenic disorders are caused by mutations of autosomal genes while about 15% are caused by mutations of sex-linked genes (X or Y). Depending on which chromosome carries the gene and the impact of mutation on its function, monogenic disorders are classified in: a) autosomal dominant, b) autosomal recessive, and, c) sex-linked. In autosomal disorders, the mutated gene is located in one of the 22 pairs of autosomes and they are characterized as dominant when the transmission of the mutated gene by one of the parents is sufficient for the development of the disease (affected phenotype)[4]. They are characterized as recessive, instead, when the mutated gene must be transmitted by both parents for the disease to develop. When a person inherits a mutation responsible for an autosomal recessive disease only from one parent, they do not manifest a clinic phenotype but they are so-called carriers of

[3] The distinction between hereditary and non-hereditary genetic disorders is critical as to the impact on the immediate family and the relatives of the person examined.

[4] Sometimes, persons carrying the same autosomal dominant mutation, present different degrees of severity; the disease has variable expressivity as they say. In other cases, the mutation has incomplete penetrance, i.e. not all the people carrying the mutation develop the disorder.

genetic disease. When two carriers of the same mutation procreate, each child has a 25% chance to inherit the mutation in question (e.g.. children with thalassaemia born by parents who are carriers of the disease).

As far as sex-linked monogenic disorders are concerned, the majority is X-linked. Males who have only one chromosome X, develop the disease if the mother was a carrier and transmitted the X chromosome that bore the mutation. In heterozygous females, sex-linked disorders occur mostly as recessive and in this case they are called carriers of sex-linked disorders.

Polygenic disorders are those hereditary diseases that are caused by mutations in more than one gene. The severity of symptoms varies depending on the number of mutated genes present in the genome, the effect of each individual mutation and their interaction.

Multifactorial disorders are polygenic disorders which develop only in combination with environmental factors. That is, the presence of mutations in more than one gene is a necessary but not sufficient condition for the development of the disease. These genes are also often described as predisposing genes. A subgroup of multifactorial disorders have a non-hereditary genetic basis because the presence of mutations is restricted to somatic cells as with several types of cancer.

Polygenic and multifactorial disorders are the most common genetic conditions and also the most difficult to understand and treat.

Mitochondrial disorders are a special category of genetic disorders associated with mtDNA mutations (mitochondrial DNA). The mitochondria are the only animal cell organelles located outside the cell nucleus that carry DNA. They are transmitted exclusively by the mother; therefore, when mitochondrial disorders are hereditary, they are of maternal origin. However, it has been observed that mitochondria may accumulate mutations during one's life span and the disease is often manifested later in life.

Finally, *diseases associated with infectious factors*, like AIDS, are not considered genetic disorders. However, genes affecting the susceptibility or resilience to the disease following exposure to the virus have been isolated.

The methods employed to determine the base sequence of particular genes or DNA regions and the methods for karyotype observation are termed genetic or cytogenetic tests respectively. Genetic tests are carried out for medical reasons: a) for diagnostic purposes, i.e. when a particular genetic "imperfection" is considered responsible for the clinical symptoms (genetic testing), b) for preventive purposes, on symptom-free persons with no previous indications pointing to a particular genetic "imperfection" (genetic screening).

Diagnostic genetic testing consists in genetic tests carried out in order to diagnose or confirm a particular disease with a known inheritance pattern, predominantly monogenic disorders. A special category of diagnostic tests are those performed on embryos.

Preimplantation tests are genetic tests performed on embryos *in vitro* when couples taking part in IVF protocols: a) have already had a child affected by a genetic disorder, b) both parents are carriers of the same monogenic disorder, and, c) there is an increased risk for chromosomal abnormalities due to the age of the mother[5]. When parents do not participate in IVF protocols but one of the above conditions is present, the genetic testing is performed on the developing embryo and is called *prenatal test*. Preimplantation diagnosis is considered by some as advantageous in comparison to prenatal testing, because in case a genetic disorder is diagnosed in the embryo, the mother is spared the psychological and physical suffering caused by an eventual termination of pregnancy. Yet, there have been concerns that preimplantation diagnosis may lead to unofficial embryo selection procedures that come very close to considerations of eugenics.

Diagnostic tests on embryos or isolated individuals are carried out on the level of clinical practice. On the level of clinical research, on the other hand, tests are carried out either on family trees or on groups of patients displaying the same clinical symptoms.

Genetic tests on family trees are envisaged when the inheritance pattern of the disease is known and there is access to biological samples of both affected and unaffected family members from different generations. In such cases, the family members' genome is analyzed with several small DNA markers scattered to all the

[5] It was found that after the age of 30 the probability of the ovum containting two chromosomes 21 (instead of the normal one) is increasing by 5% each year. This abnormality causes trisomy, i.e. the Down Syndrome, in the child.

chromosomes. Then, the inheritance pattern of the disease is correlated to the genotypes identified in these markers. In theory, this method can identify genomic regions potentially containing the gene(s) responsible for a disease. Next, these regions are analyzed in order to identify sequences potentially coding for genes. These sequences are identified for each family member and compared in order to locate the changes occurring only in affected family members.

Genetic tests on groups of persons with common clinical symptoms are envisaged either when the genetic basis of the disease or the clinical symptom in question is complex (polygenic disorders) or when genetics is only one of the components for disease development (multifactorial disorders). Precisely because in these cases there is more than one gene involved or each gene has a small contribution to the manifestation of the disease, the groups under examination must be large and researchers need to have free access to their medical records (history of disease, pharmaceutical treatment, detailed account of symptoms). DNA samples are analyzed for several markers (e.g. SNPs = single nucleotide polymorphisms) known to be polymorphic. This kind of analysis is extremely demanding if the whole genome is to be covered. Therefore, genetic testing is often focused by researchers either on genomic regions that are considered *a priori* interesting or on groups defined with strict clinical criteria. In either case, the polymorphic markers exhibiting the most significant statistical correlations with the disease are compared to the same results for these markers from a control group (unaffected persons). If no correlation is found in the control group, researchers try to isolate the genes where the specific SNPs are located or with which they are closely associated to investigate their function further.

Preventive tests may involve isolated persons or entire populations. Depending on the genetic basis of the disease under examination, preventive tests are divided in:

a) *presymptomatic tests*, when the disorder is monogenic, there is at least another family member known to be affected and the person under examination is sure to develop the disease in the future provided they live long enough and carry the mutation,

b) *carrier tests*, to assess whether a person is carrier of autosomal recessive disease or, in females, whether they are carriers of an X-linked monogenic disorder,

c) *predisposition tests*, to assess the genetic predisposition to a particular genetic disorder when predisposition is determined mainly by one gene,

d) *susceptibility tests*, to identify mutations in genes responsible for a particular polygenic or multifactorial disorder.

Preventive population genetic tests (screening) are conducted in the context of epidemiological studies in order to assess (record) the population's predisposition or susceptibility to the genetic disease in question. Such population genetic screening is important for public health policy-making.

Population genetic research is mainly conducted in the context of the human genome project which aims at recording the genetic diversity of human population. Changes of genetic makeup, in defined populations, through time and space are influenced by mutation rate, natural selection, migration and stochastic events linked to demographic size. The study of population genetic variation with respect to geographic distribution can help us comprehend the biological relationships among human populations and contributes to our understanding of human history. Additionally, the combination of genetic information with medical or epidemiological records can help identify those genetic and/or environmental predisposing factors that increase the incidence of a disease in certain population groups rather than others. The current technological advances allow the identification of the most varying DNA sequences within each population as well as among populations. Based on this variability, the genetic constitution of each population is described and then compared or correlated with ethnological, medical or other data.

D. BIOBANKS and GENETIC DATA RECORDS

The term biobank covers any private or public bank of biological samples (DNA, tissues, etc) collected either within the health care system or by related educational or research institutions. Every biological sample is accompanied by a record containing personal medical data which may be entered as words, pictures, DNA codes, antigens, antibodies or morphological, physiological or biochemical characteristics. Genetic data are part of personal data collected from genetic analysis of biological samples stored in biobanks and may cover various groups of people: a) large groups, sometimes entire populations, as in epidemiological studies, b) smaller or larger groups of patients included in clinical trials, and, c) healthy individuals used as control group.

In order to protect the confidentiality of genetic data, irrespectively of whether they form subsets of medical data or independent data sets, three basic types of records have been proposed: a) anonymous records, whereby any information able to help identify the person the data belongs to is destroyed. Anonymous records guarantee total confidentiality but have the disadvantage of being absolutely "static" since no new information can be added to them, b) encoded records, whereby personal names are replaced by numbers and the correspondence number-name is stored in a different record. These records can operate as anonymous records vis-à-vis users with no access to the link between number and name while maintaining their "dynamic" character since they can be updated, c) encrypted records, whereby all data are converted to a meaningless sequence of numbers or letters which are inaccessible without the encryption key. Encryption is mainly used to ensure access control to records which may be anonymous or encoded.

SECOND PART

ETHICAL PROBLEMS WITH REGARD TO GENETIC TESTS

In this part, the major ethical problems arising in the field of genetic tests are outlined. Many of these problems are common to different categories of tests and to different uses of their results, i.e. genetic data. In addition, the law does not treat these problems in a single way. Therefore, we thought it best to provide a "neutral", to the extent possible, account of these problems and of related views before presenting the solutions provided by different legal systems and before making specific suggestions.

1. Consent of the person concerned

I. Ensuring the consent of the person undergoing the genetic test may be considered as an essential condition with regard to any sort of genetic testing or any use of genetic data. Obviously, in the case of prenatal tests or genetic tests on cadavers, the consent cannot be sought from the person concerned but only from a relative or a close friend.

The aim of consent, in general, is to ensure personal autonomy, i.e. the ability of any person to 'self-government', free from interventions by third parties, in full awareness, according to a project of life chosen freely by the person in question. What is the place of consent in our investigation? We start from the assumption that any subject undergoing a genetic test or any subject whose genetic data are destined to some form of utilisation is in a situation of risk. The former because they may suffer physical force during removal of the biological sample; the latter because they may risk having their range of personal or social choices restricted by an abusive treatment of the revealed genetic data. In both cases, personal autonomy is threatened. For this reason, consent operates as a 'safety valve' or as the necessary and sufficient condition to protect free will or, at least, as a 'presumption' that free will was adequately protected.

Two questions arise here: a) Is consent necessary for any genetic testing or are there special circumstances that justify mandatory tests? b) In case consent is necessary, should the concerned person be fully informed about the reason of the genetic testing and the use of the results or would general information be enough?

II. The principle of prior 'free and conscious' consent of the person undergoing the genetic test following adequate information is strongly endorsed here.

One can hardly imagine anyone willing to "consent" to anything without prior information. Such "consent" would not be rational nor could it be considered as a product of free will. Yet, 'informed consent' is used as a terminus technicus both in medical ethics as well as in the doctrine of personal data protection. It refers to a more specific, stricter normative context whereby, from a moral point of view, the risk for personal autonomy is bigger as compared with other social relations also associated with the requirement of consent (i.e. economic relations, contracts, free participation in social or political organizations, etc.). This seems reasonable considering that medical tests involve interventions on the human body - the very 'material basis' of personal autonomy – and the fact that personal data - particularly 'sensitive' data, part of which are genetic data - constitute the 'core' of personality, the intrusion of which directly affects autonomy.
'Informed consent' to medical interventions is generally thought to presuppose

i) from the viewpoint of the concerned person:

a) physical capacity for autonomous will (i.e. a general capacity for understanding and deciding),

b) specific understanding of the concrete intervention,

c) independence in deciding (that is, the patient's consent is not substituted by the physician's will),

d) approval of the specific initiative suggested by the physician or having the patient taking the initiative (and giving the relevant authorization to the physician),

and,

ii) from the viewpoint of the physician:

a) disclosure of critical information, and,

b) suggestion (initiative) for an eventual intervention or treatment.

Besides, 'formal' consent is distinguished from 'substantive' informed consent. Simply filling in a consent form does not guarantee the ideal of substantive autonomous consent mainly because it depends on the particular relationship between physician/patient and, therefore, it cannot be reduced to predetermined "forms". Nevertheless, a procedure for objectively proving consent seems necessary especially to ensure the effective protection of the physician who should feel free to take every initiative required for treatment.

By way of indication, the information to be provided by the physician must include: a) elements usually considered as critical for deciding by patients, b) elements considered as critical by the physician, c) the physician's suggestion for a particular medical intervention, d) the purpose of seeking consent, e) the limited force of consent as to future interventions.

As 'interventions' on humans, genetic tests are covered by the general rule of 'free and informed consent'. This rule is established by international documents such as the UNESCO Declaration on Human Genome and Human Rights (art. 5) and – specifically for medical interventions - the Convention on Human Rights and Biomedicine of the Council of Europe (Oviedo Convention - art. 5, art. 12) and the EU Charter of Fundamental Rights, for the moment only a document of political importance (art. 3). However, some argue that genetic tests should exceptionally be

made mandatory in certain cases, e.g. monogenic[6] or incurable disease or disease requiring painful and/or unaffordable therapies. In such cases, it is suggested that older women desiring to have children should undergo mandatory prenatal genetic testing or that genetic testing should be made a prerequisite for marriage.

It is generally considered that the information to be provided to the person concerned must be complete but not to the point of manipulating the will. It is not always easy to avoid this difficulty, especially in emergencies, when patients themselves are not able to decide in time.

The extent of information is also an issue in population genetic tests carried out for medical or research purposes. It is suggested that, in this case, consent could be given as if by 'poll'. Here, of course, the information would be more general compared to individual testing.

Although it seems to facilitate things, getting consent in population tests "as if by poll" is contrary to a matter of principle: the consent of the majority cannot substitute the refusal of the minority. The overarching principle according to which the decisions of the majority stop at the boundaries of fundamental rights, i.e. the area of 'self- governance', prevails; otherwise the principle of personal autonomy would inevitably be violated. Therefore, the consent must always be strictly personal provided the person concerned is capable of understanding and deciding. In absence of such capacity, the consent must be given by the legal representative according to the general provisions of law.

Two main arguments have been employed in favor of population consent. The first refers to a matter of principle, whereby the subject of research is the population itself and thus whatever applies to individual research subjects in general should also apply to populations. In other words, no population research should be carried out without formal group consent. The second argument derives from a-matter-of-fact situation: any research on populations de facto needs to be accepted by the society it is conducted in since usually it requires the cooperation of local authorities. Population genetic research would have been impossible in the past without the

[6] For instance, thalassaemia. The genetic test identifies whether the parents are carriers of the disease. Cyprus has indirectly adopted an obligation to undergo this test (it is a necessary condition for marriage).

voluntary participation of the population in question and it often required the informal cooperation of local representatives. Sometimes the representing authority is the government sometimes religious or other leaders depending on whether the study covers the whole population or specific subsets based on historical, cultural or other criteria. At any rate, group consent should always be obtained, at least via the group's representatives. Yet, the proponents of consent in population studies recognize that, in some cases, identifying the "appropriate representatives" is difficult, if not impossible. However, in their view, international research protocols should guarantee that researchers use their best endeavours to ensure the consent of the group and that, even if they fail to identify the appropriate representatives, at least they should see that the concerned groups are consulted and their views are taken into consideration.

On the opposite side, three arguments have been raised <u>against</u> group consent. According to the first one, the lack of group consent entails an illegitimate restriction of the individual rights of the group's members. According to this view, the only acceptable restriction of the individual right to participate in a research project is the rejection of the research protocol by an Institutional Review Board. The second argument concerns the definition of the group or the population itself. The fragmentation of identity in modern societies inevitably leads to the same individual belonging to more than one group; therefore, no group can be defined unequivocally. It is thus impossible to define the "appropriate representatives", on the one hand, while, on the other hand, genetic information derived from one group necessarily contains genetic information from any other group. The third argument is based on the results of population consent. That is, while all scientists agree that genetic differences among (ethnologically defined) groups are smaller than genetic differences within each group, to seek consent only at population level not only negates this fundamental scientific finding but also enhances the role of dangerous social discrimination.

Furthermore, the distinction between tests on (a) identifiable, and, (b) anonymous samples seems to be crucial.

a) *In principle, the same form of consent is required for genetic testing on identifiable biological samples. At stake here is the protection of 'sensitive' personal data from the risk of undesired use that may restrict the range of*

personal or social choice of the person involved. Exceptionally, consent is not required when collective goods need to be protected (in particular public health[7]). However, the concerned person must be protected otherwise (e.g. by special institutional guarantees of confidentiality in the operation of bio-banks or genetic data banks).

b) *Consent is not required in case of genetic testing on anonymous samples (population tests) to the extent that personal autonomy is not threatened[8]. Therefore, different genetic testing of the anonymous samples in the future could be accepted for scientific purposes without consent. At any rate, however, any testing on anonymous samples calls for special guarantees of confidential handling of the stored samples and data files by specialized persons whenever there is a risk of objectionable use that may lead to discrimination against population groups and to subsequent restriction of personal autonomy for their members.*

It is worth mentioning that there are some generally accepted rules of self-regulation in the field of scientific research with regard to genetic tests. As to the information to be provided prior to consent, it is stressed that it should cover the aim of research, whether the collected data will be encoded in a way that allows the disclosure of the identity of the person involved or not, guarantees for the protection of privacy, whether the research results will be commercialized, what are the possible effects of participation in the research, the fact that such participation is voluntary and conscious and may be revoked at any time.

There are, also, general principles, legally established in several countries, governing DNA banks (bio-banks). According to some of these principles, the concerned person must be informed about the aims of the collection of biological samples, the risks involved in the process and the effects on other persons (i.e. family members). Written consent is required before samples can be collected or before genetic information is disclosed to third parties. Nevertheless[9], this type of formal consent for obtaining samples is not necessary when the aim of testing is obvious to the person concerned (for example, prenatal tests for hereditary abnormalities).

[7] See the relevant provisions of art. 7 (d), Act 247/1997 which are generally accepted in personal data law.
[8] See art. 7 (e), Act 2472/1997.
[9] Society for Human Genetics (Australia).

2. Medical liability

I. There is a question about the extent of medical liability, especially in case of genetic tests carried out on isolated individuals for medical purposes. Given that these tests result in the diagnosis of the genetic cause of either an already manifested disease or the predisposition for the future manifestation of disease (especially under specific environmental conditions), there is the question of whether and when should the physician recommend such tests.

II. The extent of medical liability is directly linked to the issue of prior information. Aside from the need to avoid the manipulation of the concerned person's will through such information, many people stress the duty of the physician to warn about the possibility of a false diagnosis. Besides, the physician should inform future parents of eventual treatments for diseases caused by genetic abnormality (and revealed by prenatal testing) so that they may decide on the embryo's future.

3. Eugenics

I. Prenatal testing raises the problem of eugenics: is it acceptable in IVF procedures to select specific eggs fertilized *in vitro* for transfer to the uterus (and, further, gestation) based on the results of preimplantation tests to avoid genetic abnormalities in the newborn? Furthermore, can certain genetically determined characteristics, for instance sex, be characterized as 'abnormalities' thus leading, at the end of the day, to choices guided by eugenics?

The same question is also relevant for the period after implantation in both natural and artificial reproduction: could abortion be justified for all possible 'genetic abnormalities' revealed by prenatal testing or only for some of them? Is abortion acceptable even when testing reveals external characteristics, e.g. the embryo's sex? And, finally, is there a clear criterion for distinguishing 'health' from 'eugenics', the genetically 'normal' from the genetically 'desirable'?

II. Both the UNESCO Declaration and the Oviedo Convention attempt a distinction between health protection and eugenics. Pursuant to Article 2 of the former:

'a) Everyone has a right to respect for their dignity and for their human rights regardless of their genetic characteristics. b) That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity'

Besides, pursuant to Article 14 of the Oviedo Convention:

'The use of techniques of medically assisted reproduction shall not be allowed for the purpose of choosing a future child's sex, except where serious hereditary sex-related disease is to be avoided'.

Article 3 of the EU Charter of Fundamental Rights also contains an explicit prohibition of eugenics.

Furthermore, Recommendation (90) 13 of the Council of Europe contains principles governing the conducting of prenatal tests whereby it is stressed that such tests cannot constitute prerequisites for health insurance or medical services in general.

In this context, it is argued that preimplantation tests following fertilization *in vitro* should be undertaken only when both parents have a transmissible genetic abnormality with the proviso that they do not endanger the embryo. There is also support for the view that future parents should be able to decide freely on preimplantation tests and that such tests should aim at genetic abnormalities which may cause serious disease or disease requiring early treatment. In general, it is thought that future parents (especially the mother) should maintain full responsibility for having the child, that is they should not be prevented from having the child even if in impaired health.

4. Disclosure of genetic data

I. All genetic tests raise the question of 'who' should be informed of their results. Genetic data reveal the genetic 'profile' of the person to whom they belong, they constitute a framework of basic specifications on whose basis people may plan their lives. In that respect, genetic data are part of the so-called

'sensitive data', along with data concerning national origin, political convictions, philosophical or religious beliefs or intimate privacy. Another characteristic of genetic data is that, due to the laws of heredity, they may also reveal in part the genetic identity of third persons, namely the relatives of the examined person.

Bearing all that in mind, the following questions are raised: a) Is it acceptable for persons other than the one examined and the performer of the examination (e.g. employers, insurers, banks or foundations providing grants, etc.) to have access to the collected genetic data? b) Are there any particular circumstances necessitating the disclosure of collected genetic data to third persons, for instance to prevent disease? c) Can people refuse to be informed of their own genetic data? d) Is it possible to refuse the disclosure of genetic data to their very carriers? e) What are the appropriate procedures for safeguarding confidentiality of genetic data (responsible authority, guarantee of information to the person concerned, preservation or destruction of the biological material, etc.)?

II. The basic principles governing the protection of genetic data are once again laid down in the three documents mentioned above as well as in Recommendation No R (97) 5 of the Ministerial Committee of the Council of Europe.

The UNESCO Declaration and said Recommendation are focusing specifically on this issue. The former sets out in Article 7:

'Genetic data associated with an identifiable person and stored or processed for the purposes of research or any other purpose must be held confidential in the conditions foreseen by law'.

The Recommendation makes a distinction between genetic and medical data, providing, *inter alia*, that the collection of such data is allowed for preventive, diagnostic or therapeutic purposes in the field of scientific research or in order to enable an individual to decide freely on these matters; that the collection of genetic data in civil or criminal proceedings must be stipulated explicitly by law and may not be used to identify other genetic characteristics.

The relevant provisions of the Oviedo Convention (Art. 10) and the EU Charter (Art. 8) have a broader scope. The former covers 'information about health' and the latter the 'protection of personal data'. The generally adopted principle is that everyone must have personal control of these data, therefore they cannot be excluded from such knowledge.

At any rate, the Oviedo Convention recognizes the right 'not to be informed', in case someone does not wish to know the results of the relevant testing.

The notion of the right not to know is mainly proposed with respect to preventive genetic tests, i.e. tests performed on healthy individuals and not on patients. The fact is, at least in the current context, that the majority of these tests are of small therapeutic value to the examined person. In this sense, the person under examination may choose not to know the results of the test when they feel that the resulting psychological strain is disproportionate to eventual benefits. Reported psychological adverse effects include a significant drop in self-esteem in persons diagnosed as positive or strong feelings of guilt in persons diagnosed as negative. So, people diagnosed as positive may feel guilty vis-à-vis their children because they feel they have exposed them to risk while people diagnosed as negative experience guilty feelings similar to "survivor's guilt" when other family members were diagnosed as positive.

On the other side of the right not to know lies not the right to know but the duty to know[10]. The main justification for such duty is the fact that people do not exist in isolation but decide and act within complex social and personal relationships involving others vis-à-vis whom they have moral obligations. For instance, people who are aware of being at high risk for developing a severe genetic disease because of the family's medical history must investigate it if third parties (e.g. children) depend or may depend on them in the future. Admittedly, in such cases there is a moral duty although its extent may possibly vary from person to person depending not only on character but also on the social and economic context.

Third persons may have access to these data on certain conditions, especially for reasons related to health protection, e.g. relatives who may be biologically affected by the genetic profile of a particular person due to heredity laws. It is by no means obvious, however, that access should be allowed for other reasons.

The debate on whether employers or insurers may seek the disclosure of genetic data is well known. It is generally argued that, basically, this request is not reasonable, especially when coming from employers (or potential employers)

[10] Robert Wachbroit (1966): Disowning Knowledge, Issues in Genetic Testing.

since it may lead to adverse categorizations of employees or applicants. Yet, the issue remains open with regard to professions that may contribute as environmental factors to manifestation of genetic disease - which can be avoided if the genetic data of the employee are known in time[11].

As to insurance, the argument against the disclosure of genetic data is based on the assumption that this would lead to unequal treatment (e.g. by way of oversized premiums or even by refusal to insure people genetically predisposed to serious disease). A counterargument invokes the equality of contracting parties and the overwhelming risks incurred by insurers, if they are deprived of information known to the other party. At any rate, regardless of whether insurers may ask for the results of previous tests, they may not demand the conduction of new tests because that would infringe the right 'not-to-know'. This argument seems to gain support from Article 12 of the Oviedo Convention which allows genetic testing to identify predisposition to genetic disease only for health reasons or related research. It has also been proposed to establish a certain amount above which insurers may ask the disclosure of genetic data of the party to be insured. According to another frequently proposed 'compromise' solution, this right should be limited only to specific disorders (monogenic disorders, in particular), the manifestation of which is more probable. Finally, there is the problem of 'collective insurance' (for example, trade-union members) and of the respective powers of the representing agent to negotiate the insurer's access to the genetic data of the people represented.

The procedures proposed for safeguarding the confidentiality of genetic data are similar to those concerning sensitive personal data in general. The EU Charter explicitly establishes the right of everyone to have access to their genetic data and the principle of control by an independent authority. At this point, a distinction must be drawn between the preservation of biological material and the preservation of the genetic information derived from this material. As to the biological material, according to one view, it is necessary to obtain the freely revocable consent of the concerned person for every new test other than the initial one (of course, this presupposes the preservation of the biological material).

[11] Disclosure of genetic data of employees or applicants seems justified especially when eventual manifestation of genetic disease could affect directly third persons (e.g. as in the case of Huntigton's chorea~~,~~ in ~~the case of the~~ pilot~~'s job~~).

According to another view, however, this material should be destroyed immediately after the test for which it was taken.

As to the genetic information itself, some argue that it should be preserved for a period of time exceeding one generation so that it may be used for medical purposes provided the consent has not been revoked by the concerned person. There is also support for the view that each test should be carried out for a concrete purpose, that the selected method should not reveal more genetic information than is necessary, that the tests should be performed only in specially authorized laboratories and that the person concerned should be informed of every treatment of their genetic data without having to request it.

As to the banks of biological material, it is underlined that only a limited number of authorized persons should have access to the samples or the genetic data and that these banks must develop adequate procedures for ensuring confidentiality. For prevention purposes, data banks must be organized in a way that allows them to identify persons at high risk of developing hereditary disease or whose offspring is at a similar risk.

5. Social discrimination and freedom of research

I. We already mentioned the great importance of population genetic tests for the development of research, especially for its applications in preventive medicine. However, given that these tests aim at identifying specific genetic data in populations on the basis of common characteristics (e.g. common racial origin), a problem of social 'stigmatization' may eventually emerge.

This will happen if the social characteristics of a certain group or even the personal behavior of the people belonging to it are linked to a particular confirmed genetic feature. In such cases, the members of the group are socially 'stigmatized' (either in a positive or in a negative sense), regardless of whether they participated in the particular research or not and of whether this research used anonymous samples from the group or not. This may lead to favorable or unfavorable discrimination and, eventually, to a society of objectionable biological hierarchizations. The following questions arise: should the results, even in case of anonymous samples, be protected by some degree of confidentiality? If yes, how

is free transmission of such results within the scientific community to be safeguarded to avoid impeding the progress of research?

II. Apart from the rule of anonymity of data collected for research purposes [Recommendation No R (97) 5 of the Council of Europe] (any exceptions from which are accompanied by guarantees of protection of the concerned persons), the prohibition of any discrimination based on genetic characteristics is a fundamental principle laid down in international instruments (Art. 6 of the UNESCO Declaration, Art. 11 of the Council of Europe Convention). Concerning population tests in particular, it is acknowledged that groups undergoing such tests should maintain the widest possible control. In particular, these groups should be informed beforehand of the purposes of the test; their cultural particularities should be respected as should their right to be informed of the test's results. The aim, here, is to treat these groups as equal partners and not just 'objects' of research. It is also stressed that scientific research must serve the public interest and should be proposed by a "responsible agency" [Recommendation No R (97) 5 of the Council of Europe].

THIRD PART

THE LAW OF GENETIC TESTS

Some of the ethical questions raised in the previous chapter are already answered by legislation at the level of international, EU and national law. In this chapter we will attempt a comparative presentation of the laws passed in other countries (A) and we will describe the relevant provisions of Greek law (B). Our aim is to identify the commonly accepted rules on genetic tests and the use of their results. These rules should then be taken into account when proposals for deontology are elaborated.

A. COMPARATIVE INFORMATION

1. Consent of the concerned person

At the level of international law, Article 5 (b) of the aforementioned UNESCO Declaration guarantees the principle of free and informed consent by any person undergoing genetic tests. According to the same provision consent or authorization should be obtained also in case of incapacity to consent (apparently by the legal representative of the incapacitated) guided by the concerned person's best interest.

To the extent that a genetic test for medical purposes may be considered as an 'intervention in the health field', it falls within the scope of the Oviedo Convention which is legally binding (Art. 5 combined with Art. 12). According to this document, consent must be freely revocable at any time. Furthermore, 'appropriate information as to the purpose and nature of the intervention as well as on its consequences and risks' is required before asking the consent. The same instrument establishes a framework whereby an intervention without consent is acceptable, i) when the people concerned suffer from mental disorders and risk serious damage to their health (Art. 7), and, ii) in emergency situations (Art. 8).

Both texts also guarantee the right 'not to be informed' of the results concerning the situation of health [Art. 5 (c) of UNESCO Declaration, Art. 10 (2) (b) of the Oviedo Convention].

At the level of national law, the same principles apply with some additional specific guarantees. For instance, the French Act 94-654 (Art. 22) provides that the consent of the concerned person must be given in writing in case of medical genetic tests. The abandoned 1997 US draft bill of the 'Genetic Confidentiality and Non Discrimination Act' also contained a similar term.

2. Medical Liability

Apart from the above provisions which naturally entail a corresponding medical liability, the issue of liability is wider and includes in particular: i) the principle of protection of the confidentiality of genetic data [Art. 7 of the UNESCO Declaration] which can be considered as a particular instance of

medical professional confidentiality, ii) the principle of just reparation for damaging interventions affecting the genome [Art. 8 of the UNESCO Declaration], and, iii) a general responsibility to direct research towards 'relief from suffering' and the improvement of health 'of individuals and humankind as a whole' [Art. 12 (b) of the UNESCO Declaration], on the one hand, and a duty for 'caution', 'intellectual honesty' and 'integrity' in carrying out research on the genome and in presenting and using its findings, on the other hand.

In addition, the Oviedo Convention also covers different aspects of medical liability. For instance: Article 4 refers to the compatibility of health interventions with relevant professional obligations and standards. Article 12 provides for a restricted application of genetic tests 'only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counseling', a provision which no doubt specifies a particular professional duty. The provisions of the Convention on the principles of scientific research ensuring the protection of persons involved in research (Art. 15 – 18) also affect medical liability to the extent that the advice for genetic testing can be considered as 'research'.

3. *Eugenics*

At the level of international law, this matter is also governed by the aforementioned provisions of the same documents (see above II. 3.). In Germany, the relevant law prohibits prenatal tests on undifferentiated cells. Prenatal tests with prior specific counseling to the pregnant woman are permitted, for example, under French, Austrian and Norwegian law (Act 56/1994). The latter allows preimplantation tests only for incurable diseases.

4. *Disclosure of genetic data*

In addition to the specific provisions already mentioned (Article 7 of the UNESCO Declaration and Article 10 of the Oviedo Convention), the general rules on the protection of 'sensitive' personal data also apply in this matter.

Article 6 of the Council of Europe's Convention 108/1981 'for the protection of individuals with regard to automatic processing of personal data' lays down the basic principles. The Convention requires that member-States adopt specific guarantees in addition to those applicable on personal data in general (see below in detail). At the level of EU law, there is Directive 95/46 on the protection of personal data incorporated in the Greek legal order by Act 2472/1997 which will be discussed in the relevant Section.

The special protection of genetic data can also be based on the case-law on the protection of medical data in general of the European Court of Human Rights.

Most national laws do not distinguish between medical data and genetic data. Explicit clauses on genetic data are found in article 119 (2) (f) of the Swiss Constitution (which guarantees the principle of consent), in the Austrian Act 510/1994 (Chapter IV, in particular, prohibiting the disclosure of genetic information to insurers and employers, even despite the concerned person's consent), in the 1992 Belgian Act on insurance policies (which also prohibits the disclosure of genetic information to insurers, regardless of whether favorable to the insured or not), in the 1997 Dutch Act on medical tests (which, by contrast, allows disclosure if the premium exceeds a certain limit) and in the French Act 94-653 (Art. 5, which lays down criminal sanctions in case of obtaining genetic data without the consent of their owner or in case of abuse of genetic information). The 2000 Dutch Act on protection of personal data stipulates that the processing of data for 'hereditary characteristics' is permissible without the consent of the concerned person for serious health reasons or - with special warrants of data protection - for scientific research or statistics serving the public interest.

In Australia, the general legal framework on insurance indirectly allows insurers to have access to genetic data although adverse discrimination against the insured based on these data is prohibited. In the same country, employers

may access and use genetic data only for the purpose of protecting employees from dangerous occupations[12].

At federal level, the US 1996 Health Insurance Portability and Accountability Act prohibits any discrimination based on genetic information. The prohibition is not total as it does not cover personal insurance. Many States have adopted provisions prohibiting adverse discrimination based on genetic data in insurance or even prohibiting access to such data to insurers (S. Carolina, Main, Colorado, Connecticut, Alabama, etc). In addition, 23 States have passed specific laws against genetic discrimination in employment.

With the exception of Australia[13], there are no special laws governing the setting up and operation of bio-banks for medical purposes. Such banks operate mainly on the basis of ethical rules drawn up by special scientific agencies.

5. *Social discrimination and freedom of research*

The above-mentioned Art. 6 of the UNESCO Declaration and Art. 11 of the Oviedo Convention set forth the basic rules of international law on this issue.

B. GREEK LAW

1. *Consent of the concerned person*

In Greece, the Oviedo Convention was ratified by Act 2619/1998. Thus, it is now part of national positive law and binding for any relevant legislative initiative in the future pursuant to Art. 28(1) of the Constitution. In this respect, the afore mentioned provisions of the Convention on consent are applicable here. In particular, the right of hospitalized patients to consent to diagnostic procedures (such as genetic tests) is

[12] Insurance Contracts Act 1984, sect. 21 (1).
[13] Genetic Privacy and Non-discrimination Bill 1998, n. 18.

established by Art. 47(3) of the Act 2071/1992. Paragraphs 4 and 5 of the same Article guarantee the right of these persons to prior full information. Pursuant to Art. 5 the consent is freely revocable. Besides, the Code of Medical Ethics (royal decree of 6.7.1955) establishes the physician's duty to protect personal freedom and the free will of patients in general.

2. Medical liability

The provisions of the Oviedo Convention mentioned above (see Part II, 2 II) are also applicable in Greek law. In addition, there are Article 8 (a) of the Code of Medical Ethics on medical liability and Articles 15 and 18 on the protection of medical secrecy which also covers the confidentiality of genetic data.

3. Eugenics

The above mentioned Article 14 of the Oviedo Convention applies also here. Pursuant to this article preimplantation tests are allowed strictly to avoid the transmission of a severe hereditary sex-related disease. Pursuant to Art. 304 of the Criminal Code (Act 1609/1986), abortion is permissible unconditionally until the 12th week of pregnancy (par. 4a). Until the 24th week, abortion is allowed if 'there are indications of a serious disorder which may result in the birth of a pathological child confirmed by modern means of prenatal diagnosis". The first provision seems to leave room for abortion even for purposes of eugenics while the latter covers only those cases which are characterized as 'pathological' by medicine.

4. Disclosure of genetic data

Apart from Article 10 of the Oviedo Convention pertaining specifically to the protection of personal information which also applies in Greek law and the new constitutional provision of Article 5 (5) on the 'protection of genetic identity' (related, in a sense, to the protection of genetic data), the general rules on the protection of personal data are also applicable. These include, first, the new Article 9 A of the

Constitution, the Convention 108/1981 of the Council of Europe, ratified by Act 2068/1992 and, in particular, Act 2472/1997 which implements EC Directive 95/46.
In this context, the following rules are to be underlined: a) Genetic data are part of 'sensitive data' requiring special protection in accordance with Art. 6 of the Convention 108/1981 and Art. 2 of Act 2472/1997, b) Genetic data must be collected in an appropriate and regulated manner for specific purposes, c) They must be relevant and not exceed the purpose for which they are collected, d) They must be accurate, e) They must be stored for no longer than is necessary, f) Any further processing presupposes the consent of their owner, g) Any storage of genetic data must be communicated to the Personal Data Protection Authority, h) The consent of the concerned person for the collection and processing of genetic data must be given in writing, i) Processing must be secret and necessary for medical prevention, diagnosis or treatment and may be carried out for research or scientific purposes on the additional conditions of anonymity and protection of the rights of the concerned persons (population genetic tests), j) A license for the collection and processing as well as a license for linking relevant files must be granted by the Data Protection Authority, k) The subject of genetic data has the right to be informed of their processing and eventual disclosure to third persons, the right to access the processing elements, to object to their processing and to seek temporary judicial protection.
It must also be stressed that the Personal Data Protection Authority, apart from its Opinion on genetic fingerprints in criminal procedures, adopted a Guideline on the collection and processing of genetic data of employees or applicants (115/2001), according to which the collection of such data is unacceptable even with the consent of the employee or applicant because of the unequal status of the parties in labour relations. The Guideline seems to exempt genetic data from medical reports on the condition of health of applicants specified by the general Act 1568/1985.

5. Social discrimination and freedom of research

Apart from the constitutional provisions establishing the prohibition of any discrimination (Art. 4 (1), Art. (5) (2) of the Constitution), the aforementioned Article 11 of the Oviedo Convention is also applicable on this matter. Yet, we must note that

the law on the protection of personal data does not cover data collected from samples of population tests as these data are unidentifiable.

PART FOUR

PROPOSALS FOR A CODE OF ETHICS

Based on the previous discussion and considering the main issues mentioned above, we propose the following rules of ethics.

1. Consent

a. The prerequisite of written and informed consent of the person involved should be the general rule for any genetic test. As already pointed out in our report on the specific topic of genetic fingerprints in criminal investigations, the respect of human life and dignity does not allow for mandatory genetic testing. The same rule should hold for all purposes of genetic testing.
b. The persons involved should be informed well before their consent is sought in order to ensure genuine conditions of free will. In addition, the consent should be revocable at any moment.
c. The information should cover the purpose of the test, whether the genetic data are going to be encrypted or anonymized, whether the results are going to be commercially exploited, the possible consequences of participation in the research procedure (i.e. consequences for blood relatives). More specifically, if the scope of research is to be expanded in the future, the person concerned should be informed appropriately in time to maintain the possibility of withdrawal.

2. Medical liability

a. In order to ensure free expression of will the physician must provide information on the severity of the test, its diagnostic value and its contribution to the prevention or treatment of the disease.
b. In the context of current medical ethics, the physician must safeguard the confidentiality of genetic information on condition that the health of third persons be not seriously threatened.
c. Particularly when a genetic disorder is detected in the embryo (*in vitro* or *in vivo*), the physician must inform parents about the availability or non-availability of effective treatment after birth, taking into consideration their right to reproduction.
d. The physician does not have the obligation to inform parents in case the embryo is only carrier of a genetic disease that will never be manifested.

3. Eugenics

a. For the purpose of delimitating as clearly as possible the boundaries between the protection of health and eugenics, the aim of any prenatal test (before or after the implantation of the embryo) should only be to identify a serious genetic disease, including sex-linked diseases, whose manifestation is certain or highly probable immediately after birth or during the early years of life.
b. In case preimplantation diagnosis shows some embryos to be carriers of genetic disease, they should not be eliminated from the implantation process.

4. Disclosure

a. Everyone has the right to know the results of their genetic tests but only if they so desire. However, from an ethical point of view, this knowledge seems essential when the life of a third person is affected.
b. As a rule, third parties may have access to genetic data only with the consent of the person concerned.

Exceptions from the rule of consent to disclosure of genetic data may be justified on the following grounds:

i) Protection of health of a third person.

In this case, the responsibility of disclosing the genetic data rests only with the physician.

ii) Disclosure of genetic data in labour relations

In this case the genetic data must not be disclosed. This prohibition is justified by the unequal position of employees in labour relations (especially in cases of unskilled employees or employees in areas of high unemployment).

In case working conditions may trigger the development of disease – for which there is genetic predisposition -, the employer may reasonably have access to specific genetic data with the consent of the employee when there is no alternative to eliminate or reduce the environmental risk factors.

iii) *Disclosure of genetic data in the area of insurance*

Exceptions may be justified in the area of insurance. Public or private social security funds may not ask genetic data from screenings for the same rationale described for employment (unequal position of the insured). The same holds for private insurance in absence of social security. However, any results known to the insured from previously conducted genetic tests should be disclosed. Respect of the right not-to know rules out the performance of new tests.

c. It would be appropriate to draw up a Code of Ethics (e.g. by the Hellenic Medical Association) or even to pass specific legislation on the establishment and operation of bio-banks (similar to the relevant provisions of Act 2737/1999 on Transplantation with regard to tissue and organ bio-banks). The law should clarify fundamental principles such as the possibility of access to stored samples by authorized persons and the conservation time of identifiable biological material. Act 2472/1997 covers in general the protection of genetic data.

5. <u>Social discrimination and freedom of research</u>

a. It would be useful to plan information and educational programs to familiarize the public with genetic research and to promote social dialogue on the basis of the above-mentioned principles (see Part Three, B5).

b. As to population genetic research programs, it is advisable to establish a procedure for the prior consideration and approval of research projects by specially authorized bioethics committees (Research Ethics Committees).

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