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PROPOSED INTERNATIONAL GUIDELINES

ON

ETHICAL ISSUES IN MEDICAL GENETICS

AND GENETIC SERVICES

WORLD HEALTH ORGANIZATION

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PROPOSED INTERNATIONAL GUIDELINES

ON

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AND GENETIC SERVICES

Report of a WHO Meeting on Ethical Issues in Medical Genetics

Geneva, 15-16 December 1997

Preface

The following rationale for preparing guidelines on ethical issues in medical genetics and genetic services appears in a document of the World Health Organization on Ethics and Health at the Global Level: WHO's Role and Involvement [1]:

"A great number of international committees, commissions and advisory groups and many reports and guidelines have been produced on specific biomedical issues. In dealing with ethics, WHO will not duplicate what has been done or is being done in existing national or regional forums. WHO's involvement in ethics will be directly derived from its global mission, its inclusive vision of health, and its responsibility for coordinating international health action.

Cutting across specific health issues and disciplines, WHO's main aim must be to enhance the integration of ethics in overall public health policies and practices as well as in international health cooperation. Perhaps the most pressing reason for this is to promote greater equity in access and use of health services by all individuals and in all countries. WHO will contribute to coordinating national and regional approaches, identifying gaps and workable solutions, and promoting harmonization of standards and practices at a global level."

To this end, the WHO Human Genetics Programme developed the draft document "Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetics Services" [2]. This document was circulated worldwide, and comments were received from all Regions and WHO staff. The draft document and all responses received formed the background information for a WHO Meeting on "Ethical Issues in Medical Genetics" held in Geneva from 15 to 16 December 1997. The participants at the meeting were experts in this field from both developing and developed countries.

The purpose of the meeting was to review ethical issues in medical genetics and to propose international guidelines on ethical issues in medical genetics and genetic services. These proposed guidelines, which were unanimously adopted by the participants in this meeting, appear below.

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List of Contents

-- EXECUTIVE SUMMARY --

The contents of this document were unanimously agreed upon by the experts present at the meeting. The rapid developments in the area of genetics make it necessary to continue discussions as new developments take place. For this reason, it is likely that the proposed guidelines will need modifications at certain intervals. The group felt that it is important for WHO to provide leadership in the international debate on issues related to ethics and the provision of medical genetics services.

The recommendations that follow are designed to assist decision-makers at both national and international levels to protect people and families with genetic disabilities, to recognize the great potential of advances in human and medical genetics for public health, and to develop policies and practices that will ensure that these applications can become accessible to all and are provided with due regard to ethics and justice worldwide.

The advances in human genetics that have occurred during the past twenty years have revolutionized knowledge of the role of inheritance in health and disease. We now know that our DNA determines not only the cause of catastrophic single-gene disorders, which affect millions of persons worldwide, but also predisposition to cancer, heart disease, psychiatric disorders and even to some infectious diseases.

When used properly, this knowledge will be extremely important in helping to achieve better health for people in all countries. However, these advances will only be acceptable if their application is carried out ethically, with regard to autonomy, justice, education and the beliefs and laws of each nation and community.

Genetics teaches that there is no such thing as a "superior" or "inferior" genome; humankind depends for its richness and its survival on the interaction of its complex genetic diversity with the environment.

General ethical considerations. The medical application of genetic knowledge must be carried out with due regard to the general principles of medical ethics; doing good to individuals and families, not doing harm, offering autonomy of choice after information is given, and facilitating personal and social justice. These principles, which have often been outlined by WHO and other groups, are the same as in other medical fields.

The proper use of genetic data. Since we inherit our genes from our parents, pass them on to our children, and share them with our close and distant relatives, every genetic diagnosis, test and procedure involves many people.

It is ethically imperative that genetic data should only be used to the advantage of members of a family or ethnic group, and never to stigmatize or discriminate against them.

Voluntary use of genetic screening and testing. There shall be no compulsory genetic testing of adult individuals or populations.

Every test shall be offered in such a way that individuals and families are free to refuse or accept according to their wishes and moral beliefs.

All testing should be preceded by adequate information about the purpose and possible outcomes of the test and potential choices that may arise.

Children shall only be tested when it is for the purpose of better medical care, as in the case of newborn screening when early treatment will be of benefit to the child.

Prenatal testing. Prenatal diagnosis should be offered to those who need it, but there must be no pressure on couples to accept such testing, nor to use the results of the test to compel either continuing or terminating a pregnancy when the fetus is affected with a genetic disorder.

Decisions in the context of reproduction should rest with those being tested, not with physicians or the government.

The woman should be an important decision-maker in all matters related to reproduction.

Prenatal diagnosis should be done only to give parents and physicians information about the health of the fetus; its use for paternity testing, except in cases of rape or incest, or for gender selection, apart from sex-linked disorders, is not acceptable.

Justice demands equitable access to services. Genetic services for the prevention, diagnosis and treatment of disease should be available to all, without regard to ability to pay, and should be provided first to those whose needs are greatest.

Genetic data should be treated as confidential at all times. Genetic data should only be used to advantage and empower an individual or family, and for better treatment or prevention of disease. Data relevant to health care should be collected and kept by medical geneticists in secure confidential files.

Genetic data should not be given out to insurance companies, employers, schools or governments, other than after the full informed consent of the person tested. In some countries it may be possible, or necessary, to protect both confidentiality and non-discrimination through legal means.

Genetic counselling is the provision of accurate, full and unbiased information in a caring, professional relationship that offers guidance, but allows individuals and families to come to their own decisions. Counselling is essential before any genetic testing is carried out, and should continue afterwards if the results entail choices for the person and family tested.

Genetic counselling should be available to all, and should be as non-directive as possible.

Education about genetics for the public and health care professionals is of paramount importance.

Genetics is playing an increasingly important part in medical practice, and many people are concerned about possible abuse of this new knowledge. It is important that education about genetic principles relevant to human health be emphasized appropriately for all people in all cultures.

Education is a two-way process, and geneticists and other health care professionals have much to learn from support and advocacy groups representing those with genetic disorders. Such groups are an integral part of genetic services, and should be guaranteed a voice in policy and education.

In conclusion, WHO and its constituents are urged to consider these issues as a matter of priority, to facilitate global consultation and consensus on them, and to ensure that ethical codes are put in place nationally and internationally so as to guarantee that advances in genetics can benefit the community in an ethical way. It was agreed that WHO and many other international bodies are correct in rejecting any intentional step towards human cloning as unsafe, ethically unacceptable and a distraction from the more crucial issues dealt with in these guidelines.

1. INTRODUCTION

The proposed guidelines are intended to protect people and families with genetic conditions and to inform health policy officials at the highest level of government, public health officials and workers, and physicians and other health care professionals.

The document should also be seen as a contribution to WHO's consideration of "Health Policy for the 21st Century," especially since it concerns global issues of equity in access to medical genetics services by all individuals and in all countries. One of WHO's main aims must be to further the integration of ethics in overall health policies and practices as well as in international health cooperation.

The primary purpose of these proposed guidelines is to assist policy-makers, officials, practitioners and other health workers in the Member States of WHO in ensuring that genetic information and genetic services are introduced into the broader medical practice of the nations in ethically acceptable ways. A secondary purpose is to allay fears and to reassure the public that adequate controls exist in member countries to prevent abuses of genetic information and unacceptable practices. Member States will develop their own policies and practices, using the broader principles and guidelines of this document as points of departure.

The knowledge gained from discoveries in human genetics has the potential for making a significant improvement in the health of the world's people when applied properly at the individual, family and community levels. If such discoveries and information are integrated into primary health care in ethically acceptable ways and with respect for cultural and religious diversity, the increased benefits for diagnosis, treatment and prevention of human genetic conditions will be appreciable in both developed and developing countries.

Genetics and biomedical technology open vast new avenues for research and can provide humankind with much needed therapeutic tools. But, where human life and dignity are at stake, technology cannot be left on its own to govern ethics, nor can health technology, practices and procedures be left to the vagaries of economic forces and personal interests, fears or vulnerabilities. The profound economic and technological inequalities that exist between nations and population groups have an impact on biomedical research and health care practice. Efforts must be made worldwide to mitigate such inequalities, so that the safety and rights of all individuals and communities are adequately protected and a proper balance is achieved between their rights and responsibilities.

In pursuing this task, it was recognized that some ethical problems of human genetics are highly debatable and -- at this time in history -- are issues beyond the reach of practical consensus among countries. The meeting was convened to explore the possibility of reaching moral consensus on such issues, but it could not produce consensus where this does not exist in the international community. We recognize that the laws of nations differ with respect to these particular issues, and that laws are subject to debate, evolution and change.

In the face of such controversial issues, our view is not that WHO is obliged to resolve these controversies, but that WHO can call for international unity among leaders and peoples as regards the task of including genetic knowledge and its discoveries within the goals of medicine. We recognize and respect the diversities of culture, religion and social structure that shape the public's moral attitudes in different countries. We also call for mutual respect and continued dialogue on such issues among scientifically informed members of the world's cultural and religious traditions, and for an exchange of views between developed and developing countries.

Educating the public should be a primary goal of WHO, and education is a prerequisite to informed discussion of the issues in this document. WHO should play a leading role in providing a platform for public discussion of ethics and genetics worldwide.

2. ETHICAL PRINCIPLES IN MEDICINE

The traditional sources of ethical guidelines in medicine apply also to medical genetics, which is a field of medicine (Table 1). Medical genetics' main concerns, however, extend beyond those of the traditional structure of medicine and the physician-patient relationship. For example: (a) genetic information may affect an entire family, rather than only the individual; (b) genetic discoveries may be predictive of future adverse events in an individual's or family member's health; (c) genetic information and the choices of the present may affect future generations; and (d) medical genetics has a tradition of non-directiveness in counselling.

Table 1. Relevant Ethical Principles in Medicine

Respect for the autonomy of persons: respecting the self-determination of individuals and protecting those with diminished autonomy;

Beneficence: giving highest priority to the welfare of persons and maximizing benefits to their health;

Non-maleficence: avoiding and preventing harm to persons or, at least, minimizing harm;

Justice: treating persons with fairness and equity, and distributing the benefits and burdens of health care as fairly as possible in society.

The principle of respect for autonomy includes: (a) respecting the self-determination and choices of autonomous persons, and (b) protecting persons with diminished autonomy, e.g., children and persons with mental impairments.

The principle of beneficence (L. "bene" = good) is the source of physicians' obligation to give highest loyalty to the welfare of individuals and families. Beneficence also bears upon a goal of medicine to improve the health of populations with the voluntary cooperation of the populations involved.

Non-maleficence (L. "male" = evil, harm) is the source of the traditional medical norm of "do no harm", meaning a duty to prevent harm altogether, or, if harm cannot be avoided, to minimize harm to individuals and families.

The goals of justice can be described somewhat differently: treating persons fairly, giving persons what they deserve, or giving persons that to which they are entitled. The term "distributive" (or social) justice means to allocate benefits (e.g., property) and burdens (e.g., taxation) fairly and with equity, in order to enhance social harmony and cooperation. Distributing the benefits (e.g., of diagnosis and treatment) and the burdens (e.g., of rationing of expensive care or of research risks) of health care ought to be governed by ethically justified rules such as: to each according to need, to each according to an equal share or opportunity, etc.

At present, the principles laid out in Table 1 are not applied with equal force around the world, especially respect for persons. Health professionals need to pay special attention to these principles in areas of the world where they are unfamiliar or infrequently used.

It is a common misconception that prevention and care of genetic disorders and birth defects concern only people living in industrialized countries. Genetic conditions occur with similar frequencies in different nations and irrespective of the socioeconomic status of individuals. In fact, at all levels of society, children born with genetic disadvantages have higher risks of getting sick and dying of environmental causes such as infections and malnutrition. A meaningful right to health care must include access to services for the diagnosis, treatment and prevention of genetic disorders. The priority assigned to genetic services with respect to other health services is a matter of public health policy in each country.

WHO Member States should be encouraged to draw up public health policies that include standards for genetic services along the lines recommended in the Report of a WHO Scientific Group on Control of Hereditary Diseases [3]. People have the right to equitable access to genetic services according to the standard of care that exists in each country, according to need and irrespective of the ability to pay. Also, some parties, such as women, children and people with disabilities, are especially disadvantaged and vulnerable in some societies and deserve special consideration. Professionals should help to protect such persons wherever they are at risk of harm.

Within genetic services, priority should be given to programmes that address the heaviest burdens and needs of the majority of the population. In particular, efforts should be directed towards extending the reach of genetic services at the primary care level, with the utilization of technologies and personnel that are appropriate to the needs, expectations and beliefs of the community. On the other hand, it is an inequitable use of scarce resources to develop expensive high technology services that cater only to the wealthier sectors of society while being largely inaccessible to the majority.

The principle of distributive justice should ensure that scarce resources are utilized equitably on the basis of need, and thus would oppose catering to consumers' requests for genetic services (e.g., prenatal diagnosis) to gratify cultural or personal desires rather than for medical reasons.

3. GOALS AND PRACTICES OF MEDICAL GENETICS

Medical genetics is the field of medicine that is most centrally involved in providing services to persons with genetic conditions and their families. The goals of medical genetics services are to help people with a genetic disadvantage and their families to live and reproduce as normally as possible, to make informed choices in reproductive and health matters, to assist people to obtain access to relevant medical services (diagnostic, therapeutic, rehabilitative or preventive) or social support systems, to help them adapt to their unique situation, and to become informed on relevant new developments.

Conditions studied by medical geneticists include diseases caused by defects in single genes (e.g., haemophilia, sickle cell anaemia, neurofibromatosis, cystic fibrosis), disorders caused by interaction between several genes and environmental factors (e.g., common congenital malformations, diabetes, hypertension, cardiovascular disease, breast cancer, mental disorders), and conditions caused by chromosomal anomalies (e.g., Down syndrome). Diagnostic work in medical genetics includes laboratory work at the DNA, protein and chromosome levels as well as clinical observation of disorders, including birth defects.

Whereas single-gene disorders are rare, conditions caused by an interaction between genes and environmental factors are frequent and include disorders such as cardiovascular diseases, several cancers, asthma, diabetes mellitus and mental disorders. Preventive aspects of work in medical genetics include identification of high-risk individuals with respect to common disorders for the purpose of preventing disease (e.g., heart disease) or securing early diagnosis and treatment (several cancers). At present there are significant research efforts aimed at developing somatic cell gene therapies or therapies to improve or block the function of genes.

Medical genetics services should be organized at all levels of medical care and should be directed by specially trained physicians. Actions may be conducted by a variety of health personnel according to the level of care and the particular organization of health delivery in each society. The different members of the genetics team may include Ph.D geneticists, nurses, primary care physicians, other health professionals, specially trained health care workers or genetic counsellors, social workers and laboratory personnel.

4. APPLICATION OF ETHICAL PRINCIPLES TO GENETIC SERVICES

The application of the above-mentioned ethical principles to genetic services is illustrated in Table 2.

Table 2. Ethical Principles Applied to Genetic Services

- Fair allocation of public resources to those who most need them (justice).
- Freedom of choice in all matters relevant to genetics. The woman should be an important decision-maker in reproductive matters (autonomy).
- Voluntary approach necessary in services, including approaches to testing and treatment; avoidance of coercion by government, society or physicians (autonomy).
- Respect for human diversity and for those whose views are in the minority (autonomy, non-maleficence).
- Respect for people's basic intelligence, regardless of their knowledge (autonomy).
- Education about genetics for the public, medical and other health professionals, teachers, clergy and other persons who are sources of religious information (beneficence).
- Close cooperation with patient and parent organizations, if such organizations exist (autonomy).
- Prevention of unfair discrimination or favouritism in employment, insurance or schooling based on genetic information (non-maleficence).
- Teamwork with other professionals through a network of referrals. When possible, help individuals and families to become informed members of the team (beneficence, autonomy).
- Use of non-discriminatory language that respects individuals as persons (autonomy).
- Timely provision of indicated services or follow-up treatment (non-maleficence, beneficence).
- Refraining from providing tests or procedures not medically indicated (non-maleficence).
- Providing ongoing quality control of services, including laboratory procedures (non-maleficence).

Non-discriminatory language emphasizes the personhood of those with genetic conditions. Thus, for example, someone with Down syndrome is best described as a "person

(or child) with Down syndrome" rather than a "Down syndrome child" or "Down syndrome case." Words that dehumanize persons with disabilities or stigmatize them should be avoided.

5. GENETIC COUNSELLING

Non-directive counselling has two major elements. The first is the provision of accurate, full and unbiased information that individuals and families may use in making decisions. The second is an understanding, empathic relationship that offers guidance and helps people to work towards their own decisions. In non-directive counselling, the professional avoids purposely slanting information that may lead people to do what the counsellor thinks best. Individuals and families must depend on the counsellor as a source of accurate information, and usually have no way of discovering when information is biased. Non-directive counselling does not mean presenting information and then abandoning individuals and families to make their own decisions without help. Most people may want to talk with someone who will listen to their concerns, help them to express and understand their own values, and help them to work toward their own decisions. Non-directive counsellors do not tell people what to do; decisions are those of the individuals and the families. The counsellor should, as much as possible, support all decisions.

One factor in favour of non-directive counselling was that genetics evolved as a largely diagnostic speciality with little treatment. As more treatments become available, and as susceptibility testing for common multifactorial diseases may suggest lifestyle changes that could benefit the individual's health, the counselling approach may become similar to approaches in general medicine, where the doctor may recommend beneficial treatment or lifestyle changes. Counselling related to reproductive choices should remain non-directive. The ethical principles that underlie genetic counselling and their applications are outlined in Table 3.

Table 3. Ethical Principles Applied to Genetic Counselling

- Respect for persons and families, including full disclosure, respect for people's decisions, and accurate and unbiased information (autonomy).
- Preservation of family integrity (autonomy, non-maleficence).
- Full disclosure to individuals and families of all information relevant to health. (non-maleficence, autonomy).
- Protection of the privacy of individuals and families from unjustified intrusions by employers, insurers, and schools (non-maleficence).
- Information to individuals and families about possible misuses of genetic information by institutional third parties (non-maleficence).
- Informing individuals that it is the individual's ethical duty to tell blood relatives that the relatives may be at genetic risk (non-maleficence).
- Informing individuals about the wisdom of disclosing their carrier status to a spouse/partner if children are intended, and the possibility of harmful effects on the marriage from disclosure (non-maleficence).
- Informing people of their moral duties to disclose a genetic status that may affect public safety (non-maleficence).
- Unbiased presentation of information, insofar as this is possible (autonomy).
- Non-directive approach, except when treatment is available (autonomy, beneficence).
- Children and adolescents to be involved in decisions affecting them, whenever possible (autonomy).
- Duty to re-contact if appropriate and desired (non-maleficence, beneficence, autonomy).

Full disclosure of test results includes ambiguous test results, new and controversial interpretations, and differences among professional colleagues in regard to test interpretation.

Re-contact means keeping abreast of new developments and re-contacting individuals or families on a timely basis regarding any new developments relevant to their health or reproduction, unless otherwise instructed by the individual or family.

6. GENETIC SCREENING AND TESTING

Genetic screening refers to tests offered to a population group to identify asymptomatic people at an increased risk from a particular adverse outcome. Examples are phenylalanine screening for phenylketonuria in newborn babies, or the use of maternal serum biochemical markers in pregnant women to screen for fetuses with Down syndrome. In all cases, individuals whose screens indicate that they are at higher risk must be offered a definitive diagnostic test.

Genetic testing is the analysis of the status of a particular gene. A genetic test may establish: (a) a specific diagnosis of a genetic condition in a symptomatic individual, (b) the certainty that a particular condition will develop in an individual who is asymptomatic at the time of the testing (presymptomatic diagnosis), or (c) the presence of a genetic predisposition to develop a particular complex disease such as cancer or cardiovascular disease.

The main objective of genetic screening and testing is to prevent disease or secure early diagnosis and treatment.

Ordinarily, population screening programmes are offered only when proven methods of treatment or prevention are available. In selecting population groups to be screened because they are thought to have higher than average risks, it is important to avoid the possibility of stigmatizing the entire group. Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened, in the absence of preventive or therapeutic options for the individuals screened. Screening programmes are usually better received if they work in cooperation with community leaders in the group to be screened. Screening should be preceded by educational programmes for the group.

If screening is provided for newborn babies, there is an obligation on health care providers to make sure that appropriate and timely treatment is provided. Suggested ethical guidelines for screening and testing are listed in Table 4.

A. The information provided by testing will be used to prevent harm to the person tested, or to spouse, family, prospective children or others.
B. The person is fully informed about the limitations of testing, including possibilities of uninformative results, and inability to predict exact age of onset or (sometimes) severity of symptoms.
C. The person (or the legally authorized representative) is mentally capable of giving consent.
D. Testing is accompanied by a counselling programme of appropriate length and intensity for the disorder.

In regard to requests for testing children, in the absence of medical benefit through prevention or treatment, presymptomatic or susceptibility tests for adult-onset disorders are usually best postponed until adulthood, when the young adult can make her/his own decision. In counselling, geneticists need to explain to parents the potential benefits and potential harms of testing children.

Proposed ethical guidelines for presymptomatic and susceptibility testing are presented in Table 6.

Table 6. Proposed Ethical Guidelines for Presymptomatic and Susceptibility Testing

- Genetic susceptibility testing of persons with a family history of heart disease, cancer or other common diseases of possible genetic origin should be encouraged, provided that information from the test can be used effectively for prevention or treatment (beneficence).
- All susceptibility testing should be voluntary, preceded by adequate information and based on informed consent (autonomy).
- Presymptomatic testing should be available for adults at risk who want it, even in the absence of treatment, after proper counselling and informed consent (autonomy).
- Testing of children or adolescents should be carried out only if there are potential medical benefits to the child or adolescent (autonomy, beneficence, non-maleficence).
- Employers, insurers, schools, government agencies or other institutional third parties should not be given access to test results (non-maleficence).

9. DISCLOSURE AND CONFIDENTIALITY

Disclosure and confidentiality issues are some of the most frequent ethical problems appearing in medical genetics. Because of the possibility of harm from disclosure to institutional third parties, utmost care must be taken to protect confidentiality. However, a genetic diagnosis in an individual may indicate genetic risks in his/her relatives. In those circumstances, the genetic service provider should encourage the individual to ask the relatives to seek genetic counselling. If the individual refuses, especially in cases where effective and affordable treatment or preventive measures are available, the counsellor may ethically make direct contact with the relatives, bearing in mind that the information provided should concern only their own genetic risks, not the genetic status nor the identity of the relative who refused to inform them. Counsellors should also make sure that adequate follow-up takes place.

Proposed guidelines on issues of disclosure and confidentiality are listed in Table 7.

Table 7. Proposed Ethical Guidelines concerning Disclosure and Confidentiality

- Professionals should disclose to tested individuals all test results relevant to their health or the health of a fetus. Adequate information is a prerequisite for free choice and is necessary to the open communication and trust that should mark the relationship between the provider and the person counselled.
- Test results, including normal results, should be communicated to the tested person without undue delay.
- Test results not directly relevant to health, such as non-paternity, or the sex of the fetus in the absence of X-linked disorder, may be withheld if this appears necessary to protect a vulnerable party or if prescribed by national law.
- The wish of individuals and families not to know genetic information, including test results, should be respected, except in testing of newborn babies or children for treatable conditions.
- Information that could cause grave psychological or social harm may be temporarily withheld. Within the general duty of disclosure, the counsellor may exercise judgement about when a tested person is ready to receive information.
- If a couple intends to have children, individuals should be encouraged to share genetic information with their partners.
- Where appropriate, as part of their general duty to educate, counsellors should inform people that genetic information may be useful to their relatives and may invite individuals to ask the relatives to seek genetic counselling.
- The provision of genetic information to relatives about the family so as to learn their own genetic risks should be possible, especially when a serious burden can be avoided.
- Results of carrier tests, presymptomatic tests, susceptibility tests and prenatal tests should be kept confidential from employers, health insurers, schools and government agencies. People should not be penalized or rewarded for their genetic constitutions. Information about a symptomatic condition may be disclosed as part of general medical information, in accordance with laws and practices in different countries.
- Registries (if any) should be protected by the strictest standards of confidentiality.

10. PRENATAL DIAGNOSIS

Prenatal diagnosis of genetic disorders and fetal anomalies has expanded significantly for hundreds of conditions through DNA analysis of fetal cells, and the increased use of ultrasound and maternal serum biochemical screening (amniocentesis). The purpose of prenatal diagnosis is to rule out the presence in the fetus of a particular medical condition for which the pregnancy is at an increased risk. This information is provided to the couple to assist in their decision-making process regarding the available options, such as carrying the pregnancy to term, preparing for a difficult delivery and for special newborn care, or terminating the pregnancy. Genetic counselling is particularly important prior to prenatal diagnosis and, after a result indicating an affected fetus, to secure fully informed choices. Information about the purposes, benefits and limitations of maternal serum biochemical screening must be given when offering the test, including the fact that any abnormal screening

result will need confirmatory testing by invasive prenatal diagnosis and may potentially lead to a decision about abortion.[1]

Cultures, religions and national laws differ with regard to abortion of an affected fetus after prenatal diagnosis. WHO cannot resolve these differences, but can suggest general guidelines for the provision of prenatal diagnosis, subject to the framework of the law in each individual country. Proposed ethical guidelines for the provision of prenatal diagnosis and of counselling about it are given in Tables 8 and 9.

Table 8. Proposed Ethical Guidelines for Prenatal Diagnosis

- Equitable distribution of genetics services, including prenatal diagnosis, is owed first to those with the greatest medical need, regardless of ability to pay or any other considerations (justice).
- Prenatal diagnosis should be voluntary in nature. The prospective parents should decide whether a genetic disorder warrants prenatal diagnosis or termination of a pregnancy with an affected fetus (autonomy).
- If prenatal diagnosis is medically indicated, it should be available regardless of a couple's stated views on abortion. Prenatal diagnosis may, in some cases, be used to prepare for the birth of a child with a disorder (autonomy).
- Prenatal diagnosis is carried out only to give parents and physicians information about the health of the fetus. The use of prenatal diagnosis for paternity testing, except in cases of rape or incest, or for gender selection, apart from sex-linked disorders, is not acceptable (non-maleficence).
- Prenatal diagnosis solely for relief of maternal anxiety, in the absence of medical indications, should have lower priority in the allocation of resources than prenatal diagnosis with medical indications (justice).
- Counselling should precede prenatal diagnosis (non-maleficence).
- Physicians should disclose all clinically relevant findings to the woman or couple, including the full range of variability in the manifestations of the condition under discussion (autonomy).
- The woman's and/or the couple's choices in a pregnancy with an affected fetus should be respected and protected, within the framework of the family and of the laws, culture and social structure of the country. The couple, not the health professional, should make the choice (autonomy).

Pre-test counselling makes post-test counselling (for those with an affected fetus) much less difficult because prospective parents are better prepared. Counselling should include the items in Table 9, as a minimum.

[1] References in this document to abortion as a choice available to individuals and couples following a prenatal diagnosis assumes that any such abortion is not prohibited by law. In this respect, WHO refers to the Official United Nations Report of the International Conference on Population and Development (Cairo, 5-13 September 1994) which states, in part, (para 8.25): "In no case should abortion be promoted as a method of family planning. ... Women who have unwanted pregnancies should have ready access to reliable information and compassionate counselling. Any measures or changes related to abortion within the health system can only be determined at the national or local level according to the national legislative process."

Table 9. Proposed Counselling Points prior to Prenatal Diagnosis

- Name(s) and general characteristics of the major disorder(s) that the test may identify. The list of disorders need not be exhaustive. The characteristics of the disorder(s) should be described also in terms of their effects on the future child, on the parents and on family life.
- Possibilities for treatment of the disorder(s) after birth and availability of supportive care.
- Description of the likelihood (risk) that the fetus may have the disorder(s). Risks should be expressed in several ways (as a percentage, as a proportion and verbally).
- The possibility of unfavourable test results or of fortuitous or unexpected findings.
- Alternatives available for those with an affected fetus, for example, carrying the fetus to term and caring for the child at home; placing the child in an institutional setting, if available; placing the child for adoption; termination of pregnancy; prenatal treatment of the fetus or early treatment after birth.
- The possibility of ambiguous laboratory or ultrasonography results.
- Information that, because most conditions diagnosed in the fetus cannot be treated before birth, knowing about the existence of a condition may not help the fetus.
- Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth, or professionals may not know that a family is at risk of a specific disorder (in addition to the disorder that motivated the examination).
- The medical risks to fetus and mother posed by the testing procedure.
- Non-medical risks, if any (e.g., to parental employment or health care, where applicable).
- Information that non-invasive screens used early in pregnancy, such as maternal serum alpha fetoprotein screening, may be the first step on the road to prenatal diagnosis and a possible decision about abortion.
- Costs of the test and sources of reimbursement for the mother or couple, if applicable.
- Names and addresses of genetic support groups or organizations for persons with genetic disorders that people can contact if they wish.

11. BANKED DNA

Stored DNA in tissue or blood samples may provide useful information for examination of genetic disorders in families or for research. Information from DNA specimens may be of importance for relatives and not only for the person from whom DNA originates. Therefore, access to stored DNA by family members needs to be considered.

Existing stored specimens or samples, such as those in university or hospital departments or collections of blood spots, should not be subject to new rules for consent or re-contact that may be established in the future.

In developing policies about samples to be collected in the future, it is helpful to keep the following issues in mind:

- protection of individuals from possible discrimination by employers and insurers, etc.;
- possible benefits to the individuals from research findings;
- the possibility of multiple uses of the same sample in different and unforeseen research projects;
- possible sharing of samples among collaborators, including international collaborators and commercial entities;
- advantages and disadvantages for individuals and researchers of removing all identifiers (including coded numbers) from a sample.

A blanket informed consent that would allow use of a sample for genetic research in general, including future as yet unspecified projects, appears to be the most efficient and economical approach, avoiding costly re-contact before each new research project. The consent should specify that family members may request access to a sample to learn their own genetic status but not that of the donor. While spouses may not have such a right of access, their concerns should be considered. All samples should be used with appropriate regard for confidentiality. Proposed guidelines for access to banked DNA are given in Table 10.

Table 10. Proposed Ethical Guidelines for Access to Banked DNA

- A blanket informed consent that would allow use of a sample in future projects is the most efficient approach.
- Control of DNA may be familial, not only individual. Blood relatives may have access to stored DNA for purposes of learning their own genetic status, but not for purposes of learning the donor's status.
- Family members should have access regardless of whether they contributed financially to the banking of the DNA.
- DNA should be stored as long as it could be of benefit to living or future relatives or fetuses.
- Attempts should be made to inform families, at regular intervals, of new developments in testing and treatment. Donors should inform DNA banks of current addresses for follow-up.
- After all relatives have died or all attempts to contact survivors have failed, DNA may be destroyed.
- Spouses should not have access to DNA banks without the donor's consent, but may be informed that DNA has been banked. If a couple is considering having children, it is the moral obligation of the party whose DNA has been banked to provide the spouse with any relevant information.
- Except for forensic purposes or instances when the information is directly relevant to public safety, there should be no access for institutions without the donor's consent. Insurance companies, employers, schools, government agencies and other institutional third parties that may be able to coerce consent should not be allowed access, even with the individual's consent.
- Qualified researchers should have access if identifying characteristics are removed.
- Potentially valuable specimens that could be useful to concerned families in the future should be saved and should be available.

Biomedical research in human genetics can lead to the development of diagnostic and pharmaceutical products. Patents may be necessary to raise funding to develop such products commercially, but gene sequences without proven utility should not be granted patents. Patenting has the potential to impede international collaboration, especially between developing and developed countries, to the ultimate detriment of service delivery to those with genetic disorders. Genetics differs from many areas of research in that important new knowledge can come from a family, or an ethnic group, with a particular genetic variant. If this leads to the development of a diagnostic test or new therapies, equity requires that the donors, or the community generally, should receive some benefit.

12. ASSISTED REPRODUCTION AND MEDICAL GENETICS

Although not directly related to medical genetics, various types of assisted reproduction are often discussed in connection with genetic counselling. Couples who are at risk of having a child with a genetic disorder may choose alternative options. These may include egg or sperm or embryo donation, or surrogacy. Countries have legitimate wide differences in their beliefs about the acceptability of each of these practices. In addition, these alternatives are often expensive in health resources. Whichever reproductive alternatives are offered must be consistent not only with the cultural traditions and beliefs of each country, but also with overall respect for the autonomy of individuals and families. In this context, reproductive cloning (the creation of a fetus whose genome is entirely derived from another individual) has been rejected by many international bodies, including WHO, has aroused fears in many societies, and is not in accord with currently accepted international ethical standards.

13. ACKNOWLEDGEMENTS

The participants at the meeting would like to express their thanks to those who sent their comments on the distributed draft document "Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetics Services" [2] which was used as background information for the purpose of this meeting. An updated version of the draft document, where all comments received have been considered, will be published in 1998 as a "Review of Ethical Issues in Medical Genetics and Genetic Services".

14. REFERENCES

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5. Neil A. Holtzman, Michael S. Watson, eds. Promoting Safe and Effective Genetic Testing in the United States. Report of the Joint DOE-NIH ELSI Task Force on Genetic Testing. Baltimore: Johns Hopkins University Press, 1998.

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