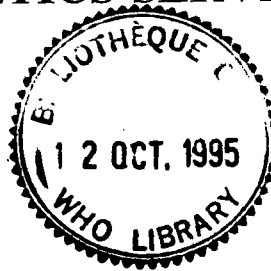


**GUIDELINES ON ETHICAL ISSUES
IN MEDICAL GENETICS
AND
THE PROVISION OF
GENETICS SERVICES**



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P R E F A C E

Worldwide, there are now over 5,000 specialists in medical genetics. At present, the majority of medical geneticists (about 3,330) work in Western developed nations, which have an overall geneticist/population ratio of approximately 1:222,000, as compared to a ratio of 1:700,000 for Eastern European nations and 1:3,700,000 for developing nations. As deaths from other causes (e.g., infant infections and malnutrition) decline in developing nations, genetics will assume a larger role. Within the next decade, newborn screening, carrier screening, and screening for common disorders such as heart disease, cancer, and neurodegenerative diseases, will greatly increase the role of genetics within medical services.

The Human Genome Initiative, while not raising generically new ethical issues in medicine, will exacerbate old ones, especially in regard to privacy, disclosure of genetic information, and freedom of reproductive choices. The Human Genome Initiative holds great promise for advances in human health, but may increase the public's fears about genetics. In order to allay these fears, to formulate rules for professional conduct, and to promote international cooperation, it is useful to develop guidelines for the provision of medical services related to genetics. Ideally, refinements to and variations on these guidelines will take place in individual nations, as a cooperative effort between genetics professionals and users of genetics services.

These guidelines promote the inclusion of clinical genetics services as an integral part of basic health care, thereby optimizing the policy of primary health care which is supported by the World Health Organization. All governments and their agencies related to delivery of health care need to examine the adequacy of the current level of genetics services and how this level can be increased by knowledge gained through the Human Genome Initiative or other research projects. New research, including that of the Human Genome Initiative, will make access to genetic information widely available on an international scale previously impossible to imagine. All individuals should have a right to know their genetic risks and risks to their potential offspring; to be educated about these risks, and to have the services available to act upon the knowledge, including the option of safe, accessible termination of pregnancies with affected fetuses if desired by prospective parents.

These guidelines therefore consider the ethical issues related to modern medical genetics and seek to demonstrate how these issues could be addressed. These approaches should be adapted at the country level, respecting cultural, economic, traditional and religious differences in societies.

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PART I : GENERAL CONSIDERATIONS

1. General Principles

Hereditary conditions affect millions of families throughout the world. About 3% of all pregnancies result in the birth of a child with a significant genetic disorder or disability. An estimated 43% of cases of severe mental retardation (IQ < 50) are caused by single genes or chromosomal abnormalities (Institute of Medicine, 1993). In developed nations, totally or partially hereditary conditions account for about 36% to 53% of paediatric hospital admissions (Andrews, et al., 1994). In developing nations hereditary conditions account for about 15% to 25% of perinatal and infant mortality (Verma and Singh, 1989; Penchaszadeh, 1993 a,b). Most non-infectious diseases, which are the major causes of death in developed nations, probably have a genetic component (Holtzman, 1989).

Even though many affected individuals may not themselves experience pain or suffering, many families remain profoundly affected by genetic conditions, in spite of improved treatment, education, and support services. In many developed nations, people with severe mental retardation and developmental disabilities now live a nearly normal lifespan. Responsibility for most of their care falls on their families. For example, in the USA, of an estimated 1 to 2 million persons with mental retardation, only about 82,000 live in institutional settings. Most of the rest live at home.

There is also a substantial cost to society in non-institutional, outpatient, educational, medical and social services, as well as lost economic output from family members who care for persons with genetic disorders.

1.1 Basic Goals of Medical Genetics

The goals of medical genetics are the diagnosis, treatment and prevention of hereditary disorders. Research is an avenue toward these goals. Prevention includes prevention of damage to genes or chromosomes arising from environmental or occupational exposures to mutagens, avoidance of conception, use of donor gametes, or prenatal diagnosis and genetic abortion, as alternatives that provide least harm. It is important that couples themselves make all decisions in these cases and that preventive measures not be imposed by medical professionals, by society, or by the law (WHO, 1989, 1993).

Good screening and counselling programmes should encourage people to take responsibility for their own health and should thus have a general health-promoting effect. The general objective of genetics services "is to help people with a genetic disadvantage to live and reproduce as normally and as responsibly as possible" (WHO, 1985). This can be done only in the context of full education and a voluntary approach to genetics services.

1.2 Facilitating Individual/Couple Choices Regarding Parenthood

1.2.1 Freedom of Choice

Promoting freedom of choice is essential to the goals of genetics. Close to 100% of 682 medical genetics services professionals¹ in a 1985 survey in 19 nations said that the following were important or essential goals of genetic counselling: (1) helping individuals/couples understand their options and the present state of medical knowledge so they can make informed decisions; (2) helping individuals/couples adjust to and cope with their genetic problems; (3) the removal or lessening of guilt or anxiety; (4) helping individuals/couples achieve their parenting goals (Wertz and Fletcher, 1989a, 1990). Clearly, respect for peoples' choices is a dominant value among genetics services professionals. This stance is laudable and justifies the spending of public health funds. In a broad sense the ability to make choices regarding one's health, including reproductive choices, may be essential to the person's integrity and contribute to psychological well-being, which is included in the WHO definition of health, namely, "a state of complete physical, medical and social well-being and not merely the absence of disease or infirmity". Therefore choice, although not the primary goal of genetics services, should be a necessary accompaniment of all genetics services. The primary goals remain diagnosis, treatment, and prevention of disease.

Freedom of choice is necessary to attain these goals. Choice has different meanings in different cultures. Professionals working in pluralistic societies need to be aware of these differences.

Choice should mean more than the absence of coercion. Choice means the practical ability to act on one's decision. In order for choice to be valid, a situation requiring choice should present more than one economically and socially viable alternative. If abortion is expensive or illegal, a woman carrying an affected fetus may have no permissible choice but to carry her child to term. If there are few services for children with disabilities, a woman carrying an affected fetus may feel that she has no real choice except abortion.

Genetics services, like other medical services, are most effective if presented in the context of an educated public that is able and willing to act voluntarily in what it regards as its own best interests. Therefore it is essential to promote public education in genetics and to protect free choice. The fact that many users of genetics services may not wish to make difficult decisions does not relieve them of this responsibility. Genetics services professionals should not place themselves in the position of making decisions for counselees² in order to lessen counsellee anxiety.

¹ The terms "genetics services professionals" and "geneticists" include M.D.'s, Ph.D.'s, and trained genetic counselors or genetic nurses in nations where these professionals exist.

² The term "consultand" is used because the term "patient" implies suffering. Many people affected by genetic conditions may not suffer. The terms "client" or "consumer" connote a business relationship and are less suitable in medicine.

1.2.2 Decision-Making in Family Context

Decisions concerning an individual's own welfare should be the province of that individual. Reproductive decisions should be the province of those who will be directly responsible for the biological and social aspects of childbearing and childrearing. Usually this means the family, which takes many forms around the world. Ideally, all persons who will support and care for a child should come to an agreement before the child is conceived or born. In cases where the various parties are unable to reach an agreement, however, the mother's wishes should be given priority.

Women have a special position as caregivers for children with disabilities. Since the bulk of care falls upon the woman, she should make the final decision among reproductive options, without coercion from her partner, her doctor, or the law.

Support for choice is based on the proposition that actions based on truly informed choices are more likely to promote human welfare than are actions based solely on laws or on professional regulations. Some people may be unaccustomed to making medical choices or may find it difficult to make such choices. Professionals should help their counselees to work toward their own decisions. Professionals should be aware of the force that the "technological imperative" (belief that availability of a procedure generates a moral imperative to use it) may exert on decision-making.

1.3 Prevention is not Eugenics

1.3.1 Medical Genetics *versus* Eugenics

Today the word eugenics usually has a negative connotation, aligned with genocide (Dunstan, 1988; Paul, 1992; Nuffield Council on Bioethics, 1993). Most professionals reject the term outright in the context of medical genetics. To most people, eugenics means a social programme imposed by the state. This is an approach to which people around the world object, because it denies human freedom, devalues some human beings, and falsely elevates the reproductive status of others.

Planned programmes can include voluntary choices. As an example of planned programmes, some nations have instituted carrier screening, on a voluntary basis and with the cooperation of the communities involved, with the expressed intention of reducing the incidence of certain severe hereditary disorders, such as beta-thalassaemia.

Individual/couple choices include avoiding conceptions, using donor gametes, or using prenatal diagnosis followed by genetic abortion to avoid the birth of an affected child. If most couples were to make the same choices, the overall outcome could be a reduced population frequency of a disorder, but it does not justify the "eugenics" label. Examples of reduced frequency of disorders resulting from individual/couple choices include dramatic reductions in incidence of Tay-Sachs disease in the USA, beta-thalassaemia in Cyprus and Sardinia, and neural tube defects in the UK (United States, 1983; Cuckle and Wald, 1984; Cão et al, 1989). In the case of neural tube defects, prevention through pre-conceptional use of folic acid may reduce but not eliminate prenatal diagnosis.

Medical genetics has as its goal the good of individuals and families. The ethos in present day medical genetics is to help people make whatever voluntary decisions are best for them in the light of their own reproductive goals. This is the decisive difference between present day medical genetics and yesterday's eugenics.

1.3.2 Voluntary Approach Necessary

Mandatory approaches, including refusal of marriage licenses, forced contraception, forced sterilization, forced prenatal diagnosis, forced abortion and forced childbearing are all affronts to human dignity. Such approaches are also bound to fail in their intended goals. In the area of reproduction, only voluntary approaches supported by the culture and by the individuals/couples involved are likely to succeed.

In undertaking genetic programmes such as carrier screening or biochemical screening in pregnancy, the primary goal must be the welfare of individuals/couples, not the welfare of the State, future generations, or the gene pool.

1.3.3 Need to Avoid Discrimination

It is important to prevent discrimination and to provide improved support services for individuals and families with genetic conditions. The absence of adequate services for people with hereditary disabilities undermines the principle of free choice for couples at risk of having children with such disabilities. In providing information to such couples, it is important to be as unbiased as possible and to avoid any actions that could be interpreted as coercive. If there is to be a reduction in the number of births of children with hereditary disorders, it is important that this be voluntary, that it be primarily for the good of the couples making the decisions, that it not detract from efforts to develop treatments for the disorders in question, and that it not result in a reduction of support services for persons with these disorders.

1.3.4 Genetic Enhancement

The remarks above apply primarily to the prevention of hereditary disorders. The genetic enhancement or improvement of "desirable" human characteristics should not be a goal of medical genetics. Geneticists should keep in mind the ethical dangers of pursuing enhancement, including increased social inequality and a lowered tolerance for human diversity. Genetic enhancement ("positive eugenics") should not be undertaken, as its consequences, intended and unintended, are not predictable at the present time. Enhancement may be difficult to resist, given our age-old desire to improve ourselves.

Enhancement violates the goals of medicine, which are (1) to relieve suffering; (2) to distribute benefits equally; and, (3) to direct research and treatment efforts toward diseases that devastate human beings. Having a modest IQ is not a disease. Enhancement is a misallocation of scarce resources.

Improving resistance to infectious disease, such as HIV, by genetic means is not enhancement, but rather the prevention of suffering, which falls within the goals of genetic medicine. It is important to remember, however, that much of what is labelled as suffering is in fact the result of social conditions or social definitions of what is normal or desirable. In the future, it may be difficult to draw a line between increased resistance to common diseases and true enhancement.

In rejecting enhancement as a good, geneticists reaffirm respect for human diversity.

2. Resources for Addressing Ethical Issues in Medical Genetics

Ethics, as a field in philosophy or religion, is concerned with systematic reflection on the moral life and its conflicts. "Ethics" is a generic term for various ways of understanding and examining the moral life and for resolving ethical problems (Beauchamp and Childress, 1994). Biomedical ethics (or bioethics) is an interdisciplinary field for the systematic study of ethical issues that arise in research, medicine and society (WHO 1992a: UNESCO, 1993). These issues can be identified within four large arenas: (1) research with human beings and animals, (2) allocation and delivery of health care resources, (3) ethical problems that arise in clinical encounters between health care professionals and patients, and (4) preventive medicine and public health.

2.1 Major Ethical Issues in Medical Genetics

This document discusses ethical problems in medical genetics today in developed and developing nations. These problems include equitable access to services, voluntary *versus* mandatory counselling, testing, and screening, safeguarding individual and parental choices, full disclosure of information, confidentiality *versus* duties to relatives at genetic risk, privacy of genetic information from institutional third parties, directive *versus* non-directive counselling, non-medical uses of prenatal diagnosis, including sex selection, and issues in research and gene therapy.

2.2 Needs of Medical Geneticists in the Study of Ethics

Belonging to a health care profession involves understanding the ethical problems that most frequently face its members in their care of patients, and in their responsibilities to society and to one another. Medical geneticists have several needs in their study of ethics:

- to know the major ethical obligations of medical geneticists in the context of the most frequent ethical problems arising in their practice today;
- to learn to lead or to participate in a process of practical moral deliberation to consider such obligations and problems, (the process must be grounded in careful examination of the circumstances of each case and respect for all persons with moral standing in the case);
- to learn to bring resources in concepts, moral experience, and professional role to bear upon such obligations and problems: e.g., (a) major ethical principles, (b) experience in prior cases and in the literature, and (c) professional values of clinicians, including caring for patients and their relationships;
- to know how to shape policies and practices to address ethical problems and prevent them, where possible.

2.3 Resources for Ethical Guidance

2.3.1 Ethical Principles

Individuals and groups must choose their sources of ethical guidance. Basic ethical principles in Table 1 (United States, 1979a; Berg and Tranøy, 1983; Fletcher et al, 1985; CIOMS, 1993; Beauchamp and Childress, 1994) are embedded in traditions and institutions in every society. Identifying and using these principles for deliberation about ethical

problems in medicine and research has been a major conceptual resource in biomedical ethics (Beauchamp and Childress, 1994). In pluralistic societies, no one ethical tradition dominates, but each tradition has a place for basic principles.

2.3.2 Knowledge and Use of Prior Cases

A second resource in ethics is knowledge of cases that, in this context, bear upon ethical problems in medical genetics. Recent moral philosophy has seen a revival of "casuistry" (Arras, 1991), a term used to refer to a method of using cases to analyze and propose solutions for moral problems. The essence of this approach is to start with paradigm cases whose conclusions are

Table 1

ETHICAL PRINCIPLES FOR MEDICAL PRACTICE AND RESEARCH

Respect for persons: the duty to respect the self-determination and choices of autonomous persons, as well as to protect persons with diminished autonomy (e.g., young children, persons with mental retardation, and those with other mental impairments). Respect for persons includes fundamental respect for the other; it should be the basis of any interaction between professional and counsellor.

Beneficence: the obligation to secure the well-being of persons by acting positively on their behalf and, moreover, to maximize the benefits that can be attained.

Nonmaleficence: the obligation to minimize harm to persons and, wherever possible, to remove the causes of harm altogether.

Proportionality: the duty, when taking actions involving risks of harm, to so balance risks and benefits that actions have the greatest chance to result in the least harm and the most benefit to persons directly involved and to members of their group.

Justice: the obligation to distribute benefits and burdens fairly, to treat equals equally, and to give reasons for differential treatment based on widely accepted criteria for just ways to distribute benefits and burdens.

settled, and then to compare and contrast the central features of these settled cases with the features of cases to be decided. To use an analogy to case law and the doctrine of precedent, when judicial decisions become authoritative, these decisions have the potential to become authoritative for other judges confronting similar cases in similar circumstances and with similar facts. Casuists hold that moral belief and knowledge evolve incrementally through reflection on cases and not from making deductions top-down from an ethical theory. Clearly, the literature in medical genetics, case reports, and anthologies of cases are valuable resources for the study of ethics.

2.3.3 Professional Values and an Ethics of Care

When medical geneticists interact with the life histories and needs of counsellors and families, ethical principles and cases are valuable but incomplete sources of guidance. Principles do orient clinicians to ethical problems, but appealing to principles does not provide a self-evident answer in the struggle to resolve a specific problem. Critics of

"principlism" in ethics hold that its language tends to focus too heavily on issues of individual rights and avoids the complex human dimensions of many bioethical dilemmas, especially issues in families and groups (Murdoch, 1970; Gilligan, 1982). Knowledge of prior cases is indispensable in making a moral judgment in a case to be decided, but no one case is exactly like another in every respect. In the final analysis, clinicians must rely on their professional values, which motivate and enable them to provide good care to patients. These values are a third resource for ethics and have been described as an "ethics of care" (Noddings, 1984; Sherwin, 1992). This view emphasizes the role of emotions and character traits in ethics. To care is to identify with other persons, each of whom is unique, aiming to nurture the web of relationships they share or can share. In settings in medical genetics, to care for a person means to identify with him or her within a plan of care that is in the best medical and personal interests of that person and that also considers the individual's relationships in families and with significant others (Berg, 1983).

2.4 The Special Position of Women and Children

Some parties are especially vulnerable and therefore need special consideration. Women usually have less favourable access to economic resources than do men (United Nations, 1991). Women may therefore suffer more than men from the effects of some decisions or disclosures, because they must depend upon the family unit for support. Women are responsible for much of the daily care of persons with disabilities of genetic origin. In making disclosures about test results (e.g., carrier status for a recessive disorder; which parent carries a balanced autosomal translocation that has caused a disorder in their child; incidental discovery of nonpaternity), and in assisting couples to reach reproductive decisions, professionals should protect the interests of those who may be vulnerable to harm from a hostile environment (WHO, 1994).

Children, persons of diminished mental capacity, and persons who may be vulnerable to harm because of their position in society (e.g., women in some cultures) need special protection from potentially adverse effects of screening, diagnostic testing, and experimentation. Professionals should serve as advocates for such persons wherever there is a possibility of harm.

2.5 Respecting those whose Views are in the Minority

Persons whose views differ from those of the majority of persons in the society are entitled to respect, even if the medical geneticist disagrees with these views. They should be treated equally with persons whose views are in the majority. For example, biochemical screening, such as maternal serum alpha-fetoprotein measurements, as well as prenatal diagnosis, should be offered equally, without regard to a woman's views on abortion. Women should also have the option to refuse the test, after full information. Although women who oppose abortion may not wish to hear about prenatal diagnosis, to withhold the offer is to treat them unequally and to prejudge their decisions. Women should be free to change their minds after testing. Couples who wish to terminate a pregnancy for what the majority regards as a minor fetal condition, or couples who wish to carry to term a pregnancy affected by what most consider a serious fetal condition should be treated equally - in terms of providing usually available services - with those who hold the majority view.

This does not mean that all technically possible services must be provided at counsellor request, but only that services normally provided be provided equally, without regard to counsellees' ethical views. Genetics services exist for the detection, prevention, and

treatment of genetic disorders. Sex selection, in the absence of an X-linked disorder, is not a medical service and does not fall under the requirement to respect minority views.

If a particular view is associated with a cultural group, one can maintain respect for the culture without accepting all practices of that culture uncritically. It is not ethnocentric to reject some practices. There are ethical imperatives that transcend cultures. For example, the United Nations Universal Declaration of Human Rights (1949) condemns slavery and other oppressive practices, even though these practices have been integral to many cultures. Sex selection, forced sterilization, forced prenatal diagnosis, and forced abortion are all oppressive practices. Even if the majority of a community, including its oppressed members, supports a practice, this does not confer ethical validity upon the practice. For example, majorities in some groups have supported harmful policies such as female circumcision.

3. Education as the Key to Ethical Genetics Services

3.1 Public Education

The goals of medical genetics can be fulfilled only in the context of an educated, informed public. Education about human reproduction and genetics should be part of the educational heritage of every person (Bankowski and Capron, 1991; Fujiki et al, 1991).

Both the principle of respect for persons and the "ethics of care" suggest that counselees should participate in decision making (see 3.2 below). Counselees are more likely to assess information accurately, more likely to reach informed decisions, and more likely to cooperate in treatment if they work together actively with professionals. In order that counselees be active participants, it is necessary that they receive some basic education about genetics.

Ideally, teaching about genetic disorders should begin in elementary school, in the context of science classes. Some basic knowledge about the hereditary nature of certain disorders that are common in a country, and about specific programmes in that country, should be provided at the elementary school level.

Most formal education about genetics will occur in high school biology courses. If biology is not a required subject for all students, information about genetics should be conveyed through other courses that may be required, such as courses on health, hygiene, family life, or general science. High school curricula on genetics should include (1) evolutionary theory; (2) how normal traits and genetic disorders are transmitted; (3) the diagnosis, treatment, and prevention of common genetic disorders; (4) special programmes for diagnosis, treatment, and prevention in a country, if any; and, (5) understanding and respecting human variability, including disability. Adult education programmes may also cover these topics, and in addition the genetics of predisposition to common diseases as well as gene-environment interaction. Educational programmes, including those for illiterate populations, must be culturally and educationally appropriate (Mano et al, 1993).

Genetics centres should be resources of information for the entire lay community, including library access and written, oral, and videotaped or filmed information at all levels. Centres should provide educational outreach to the community at large.

In providing information, educational systems should not be agents of propaganda for programmes or for directed decisions that contravene individual liberties. Public health programmes usually succeed best if people make their own informed choices.

3.2 Professional Education: A Team Approach

A team approach to genetics services is optimal to answer patient needs. In a team approach, professionals from different specialties will feel free to call upon each other to provide areas of expertise with which they are not explicitly familiar. For example, a geneticist who is not a specialist in achondroplasia may wish to enlist the assistance of a paediatrician who cares for persons with this disorder. A physician or genetic counsellor may wish to call upon a social worker to inform counsellees about financial costs and available social supports, both of which are essential elements of comprehensive counselling.

In order to facilitate a team approach, it will be necessary to provide education about genetic disorders to the persons listed in 3.2.1 to 3.2.10. Education should continue throughout a professional's career, and institutions should offer incentives to their staff to take courses or to attend meetings.

Communication between counsellee and professional is generally best if the professional is familiar with the counsellee's cultural background. Therefore it is important to include members of all cultural groups to be served in professional training programmes.

3.2.1 Physicians

Genetics and ethics should be part of all basic medical education. Physician education should include:

- Knowledge about genetics, including when to suspect the presence of a genetic disorder in a child or genetic predisposition in an adult.
- Training in counselling.
- Training in ethics. Medical education should also include instruction on how to recognize an ethical problem, as distinct from a technical problem. Ideally, courses should include exercises (often presented as case vignettes) in identifying and resolving common ethical problems.
- Teaching about human diversity and variability (WHO, 1992b). The last item is especially important for the practice of medicine generally. Genetics plays an important role in individuals' variable responses to infection, degenerative diseases, medication, and diet. Courses should include conditions of wholly or partially genetic origin affecting both children and adults. Genetic contributions to common diseases, such as heart disease, cancer, or diabetes should be part of medical education.

3.2.2 Nurses

The role of nurses in taking family histories and providing genetic information and counselling will become increasingly important in the future. Genetics should be part of all basic entry-level programmes for the training of nurses. Advanced programmes for training nurse-specialists in genetics should be encouraged.

3.2.3 Midwives

Midwives need education about inherited disorders and available prenatal screening or testing.

3.2.4 Genetic Counsellors

In a few nations, notably the United States and Canada, genetic counselling exists as a separate profession. Counsellors have received advanced post-graduate training in both genetics and psychosocial counselling, but are not physicians. The counsellors' training is uniquely suited to counselees' needs because of its emphasis on counselling. Counsellors' training is less lengthy and less costly than training physicians. Establishment of genetic counselling as a profession should be encouraged in all nations.

3.2.5 Single-Gene Counsellors

It may also be appropriate to train laypersons to counsel for some single-gene disorders that are common in a particular population. Training can be focused and cost-effective. Single-gene counsellors have worked effectively for families affected by sickle-cell anemia in the USA.

3.2.6 Social Workers

Social workers are often the liaison for social and financial support services and for reimbursement of medical services. They often provide counselling and therapy for individuals and families. They need sufficient education about genetic disorders in their basic training to provide individuals and families with optimum service.

3.2.7 Community Health Workers

Community health workers, including rural health workers, who provide much prenatal and postnatal care, need education in family history-taking and newborn screening. In some rural areas where they will be the ones to deliver test results, they should also receive basic training in supportive counselling skills.

3.2.8 Pharmacists

Sometimes genetic conditions may affect responses to drugs. Therefore, it is important that pharmacists as well as pharmacologists be aware of these differing responses and be prepared to suggest substituting alternative medications if necessary.

3.2.9 Nutritionists

Genetic conditions may affect uptake and metabolism of dietary nutrients. In turn, some genetic conditions can be treated by diet. Nutritionists are often responsible for overseeing dietary treatments and may sometimes contribute to diagnoses. They need education about genetic conditions.

3.2.10 Oral Health Professionals

Dentists and oral surgeons should receive education about those genetic conditions affecting the mouth and facial structures.

3.3 Others Associated with Service Provision

Many other personnel play important roles in patient care or provision of services. Their quality of performance can mean the difference between life and death. These persons include suppliers of medical equipment for home care (e.g., oxygen, kidney dialysis supplies, physical therapy equipment), respiratory therapists, home health aides who assist in tasks of daily living, and medical office managers who schedule appointments, take initial counsellor information, keep records, and deal with bills. These personnel should not be regarded as merely peripheral to patient care. In some cases, such personnel will have extensive on-going contact with counsellees and may even, perhaps inadvertently, make decisions that affect the counsellees' health. It is important that such persons receive some education about common genetic disorders so that they can better communicate with counsellees. It is also important that they receive instruction on the ethics of confidentiality and disclosure.

3.4 Clergy

Clergy officiate at over 80% of weddings in some parts of the world (Fletcher, 1982). They are therefore in a unique position to sensitize couples and their extended families to the potential effects of genetic disorders before a betrothal or wedding takes place. In some nations, clergy may also act as supportive counsellors as couples work through the decision-making process related to handling genetic information. At present, clergy are not given adequate education about genetic disorders.

3.5 Organizations for Affected Families

Organized groups of individuals and families affected by genetic disorders exist in many nations. Members of these groups can help to educate the public and can provide information about their experiences to those recently diagnosed. These groups can be one of the best sources of practical help to families engaged in daily care and education. They can also keep individuals, couples, and families abreast of new developments in diagnosis and treatment. Family-to-family communication should be an essential part of the genetic counselling process. It is important that patient organizations be kept informed about all developments in diagnosis, treatment, and research so that they can continue to inform their memberships. Organizations for persons affected by genetic disorders should work with professionals as an integral part of a team for counsellee education and care.

3.6 Preventing Stigmatization

Education has the potential power to prevent stigmatization and discrimination by emphasizing that genetic disorders are not caused by the behaviour of affected persons or families (Nuffield Council on Bioethics, 1993). Education can be an equalizing force. Education should stress the point that most people may carry some recessive lethal mutations and that we or our offspring are all at genetic risk.

4. The Contexts of Genetics Services in Health Care Systems

Genetics services should be provided in the contexts of premarital health visits, family planning, preconceptional care, prenatal care, pediatric and adolescent care, and adult care. This list is illustrative of potential services, but is not exhaustive. This approach is in line with the concept of primary health care (PHC) which has been developed by the World Health Organization. The core principles of PHC are concerned with equity, efficacy,

effectiveness, community participation, and providing possibilities for the improvement of health and well-being of populations (WHO, 1990). Basic guidelines are illustrated in Table 2.

The team approach described under 3.2 above is optimal in all contexts.

Table 2

BASIC GUIDELINES FOR GENETICS SERVICES

1. Goals: help people with a genetic disadvantage live and reproduce as normally as possible.
2. Equal and fair allocation of public resources to those who most need them.
3. Freedom of choice; woman should be final decision-maker in reproductive matters.
4. Voluntary approach necessary; avoid coercion by government, society, or physicians.
5. Respect for human diversity, and those whose views are in minority.
6. Respect for counselees' basic intelligence, regardless of knowledge.
7. Education about genetics for public, medical professionals, teachers, clergy.
8. Close cooperation with organizations for persons with genetic disorders, if such organizations exist.
9. Prevention of unfair discrimination or favouritism in employment, insurance, or schooling based on genetics.
10. Teamwork with other professionals through network of referrals. When possible, help counselees become informed members of the team.
11. Use of nondiscriminatory language that respects individuals as persons.

4.1 Premarital Genetic Counselling

Cultures differ widely in their traditions of gender roles, marriage, parenthood, and family life. However, in spite of such diversity, one of the most universal values among persons and their communities is expressed in the hope of having healthy children. All may share in this hope, but not all share in the opportunity to minimize the dangers and burdens of heredity to children. Ideally, wherever genetic counselling and testing exist, this opportunity can be pursued well before couples engage in decision making about marriage. Such counselling and testing should be at the will of individuals, couples and communities; governments should not require premarital genetic counselling or testing by law. Laws requiring such counselling or testing would violate the principle and practice of voluntariness in genetics services. Voluntariness is the greatest safeguard against misguided returns to the eugenic thought of the past.

4.1.1 Choice of Partner

In cultures where arranged marriages are the norm, premarital testing for recessive disorders common in particular populations may avert unions at high genetic risk. In order to prevent stigmatization of individuals or families, it is important that test results be kept strictly confidential. Each individual involved (that is, the members of the prospective couple) should have full knowledge of the test results, together with full education and supportive counselling. It may be necessary to educate other family members in order to prevent prejudice against carriers.

Prospective carrier-carrier couples whose marriages are not arranged should receive full information and counselling about reproductive alternatives with both parties present, as described under 4.1.2. Geneticists should recognize, however, that in many communities marriage serves other social and individual purposes in addition to reproduction, and that many couples will not base marital decisions on genetic information.

4.1.2 Reproductive Alternatives

Alternatives for couples at genetic risk include not having children, taking their chances of having a child with a genetic condition, having prenatal diagnosis and either selectively aborting affected fetuses or carrying them to term, adopting a child without the disorder, the use of donor gametes, and/or other methods of assisted reproduction. (Also for discussion and guidance on issues in this section, see Council of Europe, 1993; Royal Commission on New Reproductive Technologies, 1993).

Adoption

Adoption, including cross-cultural adoption, should be encouraged as a means of family formation. Adoption is a time-honoured means of forming a family and should be regarded equally with biological means of family formation. There should be as few restrictions as possible for prospective adoptive parents. The adoption process should be affordable to all; third parties should not profit economically from adoption.

Many couples, however, will still want to have biological children. Their wishes should be respected.

Donor Insemination

Donor insemination should be available, affordable, and legal everywhere. Donors should be screened by way of a family history of genetic disorders and should be tested if appropriate. They should also be tested for HIV infection and other sexually transmitted diseases. In order to prevent marriages between close biological relatives in future generations, there should be a limit on the number of donations from a given individual.

Egg Donation

Egg donation places considerably more burden, in terms of time, risk, and discomfort, on the donor than does sperm donation. The burdens are due to the need to stimulate the ovary and aspirate ova ripe for fertilization. In the best hands, there is a modest risk of infection (1%) due to the aspiration procedure, and of hyperstimulation syndrome (<1%) from use of fertility drugs. A third risk, which is unproven, is an association with ovarian cancer from use of fertility drugs. Egg donation also requires in-vitro fertilization (IVF) or

gamete intrafallopian transfer, which is costly. There are some potential benefits from use of egg donation, namely that both partners can regard the child as biologically theirs, the father because he provided the sperm and the mother because she carried the child. These benefits, however, may not outweigh the risks and costs of IVF. Therefore egg donation should not be encouraged if donor insemination would avoid the genetic problem (e.g., for autosomal recessive disorders). In situations involving X-linked disorders or autosomal dominant disorders in the woman, egg donation may be appropriate. Because of the extensive discomfort and time considerations, egg donors should be compensated for their time, effort, and lost wages, but there should be a limit on the number of pregnancies accomplished by donations from one individual, both to protect the woman's health and to prevent potential consanguinity in the next generation.

Embryo Donation

IVF may result in the fertilization of more embryos than are needed for implantation. Usually these "surplus" embryos are frozen and stored for possible further IVF attempts by the parents. If the woman does not need or wish to use the frozen embryos, however, she and her partner may choose to donate the embryos for use by a woman or couple at genetic risk. Use of a donated embryo is one way to avoid the risk of an affected child. Only surplus embryos accidentally resulting from IVF should be used; embryos should not be created solely for purposes of donation. All donations should be voluntary; donors should not receive compensation, and embryos should not be bought or sold.

Surrogate Motherhood

Surrogate motherhood takes various forms in different cultures. Some of these are highly exploitative of women. Surrogacy has the potential to harm both the biological and the social mother. IVF with a donor egg has a lower success rate in terms of live birth than does surrogacy, but it is still to be preferred. Any form of surrogacy that uses the biological mother against her will or employs economic coercion is akin to slavery and should be condemned.

Disclosure of Biological Parentage

Children who are adopted or who are conceived from donor gametes should be able to find out the names of their biological parents, on attaining legal majority, if and only if the parent(s) have consented to be found. Discovery should be mutual. This end is best achieved by establishing consensual registers of donors or birth parents to whom disclosure is acceptable. These registries should be periodically updated and registrants should have the option of removing their names. Children should be provided with a genetic health history of their biological parents even if names are not revealed.

4.2 Family Planning

Genetics services should be included in larger family planning programmes that present couples with the full range of options described above, including full information about contraception.

4.2.1 Family History

Couples intending to have children should be encouraged to meet with their physician, nurse, midwife, or other professional before conception in order to examine their family

histories and to discuss other risks such as advanced maternal age, family history of genetic disorders, or environmental or occupational exposure.

4.2.2 Carrier Testing

Carrier testing in high-risk families or populations (e.g., for Tay-Sachs, sickle-cell, or beta-thalassaemia) should preferably be performed before rather than after conception, because it allows a choice of preconception alternatives. All testing should be voluntary. Couples should be fully informed.

4.2.3 Counselling

In cases where a couple's risk significantly exceeds population risk, discussions should include a full and unbiased description of how someone with the disorder in question develops over the entire life course. Such descriptions should include the full range of variability of the disorder, effectiveness of education and treatment, and availability of prenatal diagnosis if applicable. It is not appropriate to tell mentally competent couples that they should not have children. This should be their decision, on the basis of full and unbiased information. The only exception to this rule is a situation where pregnancy, labour and delivery threatens the mother's life or long-term health. In such cases the professional may argue against initiating pregnancy, but the final decision should be the woman's. In assessing a couple's competence, professionals should use standard criteria ordinarily employed in other medical decisions. These include (1) evidence that the individual's actions are voluntary; (2) "reasonable outcome" of a choice in terms of the individual's and family's social and cultural situation; (3) sound reasons for the choice; and, (4) understanding of risks, benefits, and alternatives, including knowledge of both facts and implications.

Genetic risks associated with advanced maternal and paternal age should be explained. Couples should be encouraged to complete their childbearing before the ages of highest risk if the alternatives of prenatal diagnosis and genetic abortion are not acceptable or available. Professionals should take care, however, not to impose their own values on couples' lives. Some couples may prefer to assume the risks of having children at later ages rather than reorganize their life plans around genetic risks.

If a couple plans to have children, appropriate dietary measures (e.g., folic acid supplements to prevent neural tube defects or a strict low phenylalanine diet for women with phenylketonuria [PKU]) should be instituted in special cases before conception. Such diets should be supported with public funds.

4.2.4 Parenthood for Persons with Disabilities

Many people with disabilities can bear and raise children successfully if they have sufficient support. Professionals should be supportive of their desire for children and counselling should include a full description of the implications of parenthood for parents, child, and the family, including the probability of transmitting a parental disorder to the child. In cases where the disorder may become more serious in succeeding generations (e.g., myotonic dystrophy, fragile-X syndrome), the counsellor should make clear the risks of biological parenting and should discuss other options. Disorders associated with possible expansion of a genetic error are morally troubling because of risks of increasing disability in children and grandchildren.

Parents everywhere, including those with disabilities, desire children with a reasonable expectation of leading a healthy life. A society can show its respect for human diversity and its solidarity with parents by respecting their desires for children and by helping them to foresee how an affected child can be helped to participate in society alongside other children in the child's birth cohort. Persons with disabilities, including blindness and deafness, should not be excluded from adopting children, provided they can care for a child.

4.3 Preconception Care

Preconception genetics services for couples intending to conceive in the near future should occur in a larger context of pre-conception care (United States, 1989) that includes the following:

Preconception Risk Assessment

Preconception risk assessment offers the opportunity to identify:

- individual and social conditions, e.g., extreme obesity; advanced maternal age; special diets; and vocational, housing, and economic status; physical abuse;
- adverse health behaviours, e.g., use of tobacco, alcohol, and illicit drugs;
- medical conditions, e.g., immunity status, medications taken, genetic status, acute and chronic illness;
- psychological conditions, e.g., stress, anxiety, and depression;
- environmental conditions, e.g., workplace hazards, toxic chemicals, radiation; and
- barriers to family planning or early prenatal care enrolment.

Health Promotion

Preconception health promotion offers the opportunity to provide:

- counselling about safer sex, pregnancy planning, spacing, and contraception;
- counselling about the availability of social programmes;
- advice regarding over-the-counter medications; and
- information on environmental and occupational hazards.

Intervention to Reduce Medical and Psychosocial Risk

The preconception visit provides an opportunity to intervene in medical or psychosocial risk identified by risk assessment. Such intervention may include:

- treatment of maternal and paternal disease identified, including infections;
- modification of chronic disease medication and regimens to decrease teratogenic risk;

- carrier testing for persons with a family history of genetic disorder and members of high-risk ethnic groups;
- vaccination;
- counselling regarding behaviours, including those related to HIV and other infections;
- nutrition counselling, supplementation, or referral;
- substance abuse counselling or referral to treatment programmes;
- home visiting to treat psychosocial risks;
- provision of social services and financial assistance;
- discussion of the importance of early prenatal care;
- referral to other health care providers, e.g., community mental health centre;
- discussion of alternative options, such as use of donor gametes, if a couple is at genetic risk;
- provision of contraception or referral for family planning.
- psychological, social, and financial preparation for the birth of a child with a genetic disorder;
- referral to organizations for families affected by genetic disorders (support groups);
- information about prenatal tests, where applicable.

Preconception Care Delivery

Practitioners should give information regarding future childbearing as part of routine health maintenance. In addition, preconception care in primary care practice can be included in visits for other purposes: the school physical examination, the premarital examination, the family planning visit, and well-child care for another member of the family.

4.4 Prenatal Care

Genetics services should be an integral part of prenatal care. The overall content of prenatal care should include social risk assessments and health promotion activities, as described above under Pre-Conception Care. Family genetic history should be taken at the first pregnancy visit if a preconception visit has not taken place.

Carrier testing should be offered to persons with a family history of a genetic disorder for which testing is available and to members of ethnic groups at elevated risk. Carrier testing should be voluntary. Ideally, before testing, both members of a couple should be fully informed about their genetic risk and about the medical, social, and economic aspects of the disorder in question. For disorders of variable expressivity, the full range of manifestations, from minimal to severe, should be presented. Full pre-test information should include descriptions of how the disorder affects development over the entire course of the life cycle.

Full pre-test information should also include a description of available options, such as prenatal diagnosis and abortion of an affected fetus, in case both partners are carriers. Although discussion of prenatal diagnosis may seem premature, it is generally best to inform people, before the initial carrier test, that the carrier test may lead to a difficult decision. Early information allows time to prepare oneself psychologically for a possible adverse outcome or to refrain from the test.

The ideal provision of information by professionals may not always be possible, in view of limited resources. Trained laypersons, single-gene counsellors, written materials, movies, and videotapes could supplement and in some cases substitute for professionals. Special attention should be given to methods of providing basic pre-test information to illiterate or semi-literate persons and to obtaining their informed consent. If resources are limited, efforts should be concentrated on persons with the highest risks.

Prenatal tests should be offered if medically indicated (See Part 2: 12, 13 below). Women who enter prenatal care too late for prenatal diagnosis should receive information about it in order to encourage them to seek prenatal care earlier in their next pregnancy.

Refusal or acceptance of a carrier or prenatal test should be the individual's or the couple's choice and should not affect their medical care or their child's medical care in any way.

4.5 Childhood Care

Today most patients bringing a couple in contact with a genetics clinic are children under the age of 14. The physician should explain to parents that they are not responsible, in the sense of being culpable, for the child's disorder. In interactions with parents of children with genetic disorders, the doctor should regard them equally with the parents of "normal" children. The physician should explain to both parents that their actions did not cause a genetic disorder, and should explain to the mother that her behaviour before or during pregnancy did not cause the child's genetic condition. It is important that this information also reach the father, lest he blame her.

In interacting with the child, emphasis should be on the child as a person rather than as a bearer of a genetic condition. The physician should use the same approach as would be used with children without the genetic condition, insofar as possible. Parents should be encouraged to raise the child so that the child will have normal self-esteem.

Whenever possible, children should be informed about their condition and its treatments and should be given an opportunity to discuss the treatment. From approximately the age of seven, mentally competent children will often understand a basic description of treatment alternatives. As the child matures, greater weight should be given to the child's wishes. Cultures vary in terms of the ages at which they consider a child capable of making decisions about the future. Many of the world's religions regard the ages of 11 to 12 as the age of discretion. Treatment generally proceeds with less difficulty if the child or adolescent is a willing and informed participant.

As children with genetic disorders reach adulthood, it is important that there be a smooth transition from pediatric to adult care. In cases where survival to adulthood is rare and a pediatric clinic may be the only source of care for an adult, clinic staff should make every effort to respect the psychosocial needs of adult patients.

4.6 Adult Care

In the future, the majority of genetics services may be provided to asymptomatic adults seeking to learn their risk of developing heart disease, cancer, diabetes, mental disorders, Alzheimer disease, or other common diseases (Berg, 1994). Testing, and eventually genetic treatment for those who are susceptible to these disorders will become part of routine adult care. Genetics will shift from a specialty related to pediatrics or obstetrics to an adult specialty closely allied to general practice and adult-type preventive medicine. Genetic risk testing could become part of routine physical examinations.

5. Priority of Genetics Services in Health Care Systems

5.1 Distributive Justice

The two-sided problem of access and inadequate services is the most significant social-ethical issue in human genetics today. The basic issue is one of distributive justice, especially when a society can provide fairer access for those at higher genetic risk and can increase services to meet the need but does not act to do so. In ethics, "ought implies can." Ladd (1973) argued that this maxim points to a presupposition of moral discourse itself. If persons, groups, or societies "ought" to do something but "cannot," then "the moral proposition containing the ought is void and pointless." The maxim "ought implies can" clearly bears upon a society's obligation to distribute health care resources fairly, and to be fair when distributing genetics services. A few societies have the economic, professional, and technical resources to approximate or reach the population's level of need for genetics services. Many more societies do not have an assembly of such resources today, but they have the ability to engage in a long-term process of developing the resources necessary to assign an appropriate priority for genetics services among the other needs in health care of a population. Still other societies are so beset by conditions of war, famine, poverty, and geographical isolation that their capacity to respond to all basic health problems, including those related to medical genetics, is severely limited. No moral judgment should be assigned in such instances, because the capacity to act is not present.

In setting priorities for genetics services, it is important to remember that the majority of infant and young adult deaths on a worldwide basis have non-genetic causes: poverty, infection, malnutrition, violence, lack of basic medical care. These problems must be resolved. It is unjust to provide high-technology services to a few who can afford them, while failing to provide basic care to the majority.

The principle of justice requires that services should not be rationed on the basis of ability to pay. A national health care system that provides essential care for all, regardless of ability to pay, is the most ethical approach. Genetics services, including newborn screening, carrier testing, providing special diets, such as the PKU diet, prenatal diagnosis, abortion of affected fetuses, and treatment should be included in national health care systems. Since resources are not infinite, priorities for the provision of services should be determined on a basis agreed upon by the communities to be served.

Access to genetics services should be distributed equally across a country. Clinics should include regular outreach to rural areas whenever appropriate.

5.2 Cost-Benefit Considerations

Cost-benefit analyses, when required in setting priorities for public health programmes, can be held to the following ethical standards:

- Cost-benefit analyses should be as realistic as possible in terms of families' lived experiences. (It makes no sense, for example, to assume that most parents will raise a child to the age of 18 and then turn the child over to a residential institution for life.) In the interests of improving accuracy and eliminating unfounded assumptions, those planning cost-benefit analyses should include representatives of organizations for persons with genetic disorders and affected individuals or family members as integral members of the project team.
- Cost-benefit considerations should not be used to establish arbitrary limits on genetics services, e.g., limiting the length and number of genetic counselling sessions without regard for the needs of individuals. This is especially important for services such as counselling, where results may not be quantifiable.
- Cost-benefit analyses should include non-monetary costs and non-monetary benefits in their calculations. For example, not having children is an emotional and social cost for most families, while having healthy children is a benefit. Genetic abortion of any pregnancy is an emotional cost, while relief from anxiety after favourable prenatal diagnostic results is a benefit. Cost-benefit analyses should include the non-monetary costs and non-monetary benefits of a programme, including psychological and social costs and benefits to individuals and families. Policy makers should weigh these costs and benefits in making their decisions.
- There are ethical problems inherent in the very idea of cost-benefit analyses. A fundamental limitation of the cost-benefit approach is that costs (of whatever kind) often accrue to one sector of society and benefits to another. All cost-benefit analyses should include a statement on the ethical and social limitations of the analysis and on potential harms that may arise from these limitations. These have been summarized as follows:

"Uses and Limits of Cost-Benefit Analysis. Cost-benefit analysis has become a recognized tool for making allocational decisions in a broad range of areas, including health care. It can help answer resource allocation and access questions concerning genetic screening and counselling, provided the significant limitations of the method are clearly understood.

Cost-benefit analysis is most useful when the costs and benefits of the action under consideration are tangible, can be measured by a common unit of management, and can be known with certainty. These conditions are rarely satisfied in public policy situations and they can be particularly elusive in genetic screening and counselling programmes. For example, cost-benefit calculations can accurately evaluate the worth of a projected prenatal screening programme if the only costs measured are the financial outlays (that is, administering a screening and counselling programme and performing abortions when defects are detected) and the benefits measured are the [funds] that would have been spent on care of affected children. But the calculations become both much more complex and much less accurate if an attempt is made to quantify the psychological "costs" and "benefits" to screenees, their families, and society.

A more fundamental limitation on cost-benefit analysis is that in its simplest form it assumes that the governing moral value is to maximize the general welfare (utilitarianism). Simply aggregating gains and losses across all the individuals affected omits considerations of equity or fairness. Indeed, cost-benefit methodology itself does not distinguish as to whose costs and benefits are to be considered. But in the case at hand, it is an ethical question as to whether the costs and benefits to the fetus are to be considered, and, if so, whether they are to be given the same weight as those of the mother and family.

It is possible, however, to incorporate consideration of equity or fairness and thereby depart from a strictly utilitarian form of cost-benefit analysis either by weighing some costs or benefits or by restricting the class of individuals who will be included in the calculation. In any case, cost-benefit analysis must be regarded as a technical instrument to be used within an ethical framework (whether utilitarian or otherwise), rather than as a method of avoiding difficult ethical judgements.

In general, the process of attempting to ascertain the costs and benefits of a given policy according to a common standard of measurement performs the useful function of forcing policy-makers to envision as clearly as possible the consequences of a decision. For example, the health authorities in cities with few marriages between Ashkenazi Jews might decide not to mount a Tay-Sachs screening programme on the ground that the rarity of the expected occurrence would raise the cost-per-case-detected to a very high level in light of the expected savings. Yet their ethical analysis will need to recognize that the risk of a Tay-Sachs birth for an individual Ashkenazi couple is the same whether the benefits and burdens are distributed fairly or not.

More particularly, cost-benefit analysis can rule out some policy proposals, once ethical priorities have been fixed" (United States, 1983; Modell and Kuliev, 1991).

For example, the benefits of the knowledge gained through screening of elementary school children may not outweigh the administrative costs and the possible social stigma that could be suffered by those screened.

5.3 The Role of Users of Genetics Services in Establishing Policy: Need for Grievance Procedures

Counsellors have a special perspective on genetics services that should be integral to policy and planning. Users of genetics services, including adults with genetic disorders as well as parents of children with genetic disorders, should be on the boards of genetic testing centres.

With the help of counsellors, genetics centres should establish procedures for reviewing complaints and should inform all counsellors of the existence and location of the office or person to whom they may refer complaints. A review board that includes both geneticists and counsellors and that has investigative and enforcement powers is optimal. Existence of grievance procedures in the long run helps to improve services.

PART II : GUIDELINES FOR SPECIFIC SERVICES

1. Basic Principles in Professional-Counselee Interactions

Both medical geneticists and counselees have responsibilities. Several of these are outlined in Table 3.

1.1 Respect for Persons

Respect for persons should be the basis of all genetics services (Table 4). This is not only the most ethical approach, but also the most effective in terms of communication and care. Geneticists should regard counselees as partners in their own care. This means respecting counselees' intelligence, whatever their level of education. It means listening to counselees and letting them talk without interruption. Studies have shown that this is the most effective means of obtaining important medical information (Beckman and Frankel, 1984). Counselees' comments and questions should be taken seriously. Although a question may reflect a lack of basic knowledge about genetics, this does not mean that the individual is unintelligent. The person's question has meaning for that person and deserves a serious answer. Ideally, the professional should try to gauge a person's knowledge at the outset of the session, by asking people to describe their perception of the situation, so that the counsellor can adjust the level of language to the counselee's level of current understanding. The approach avoids making already knowledgeable counselees feel belittled by presentation of basic information with which they are already familiar. Although some may lack formal knowledge, they are intimately aware of their own bodies and of their family members' day-to-day symptoms. Their experience gives them claim to a kind of expertise about their conditions.

Most counselees need emotional support. The supportive aspect of counselling is of at least equal importance with the informational respect. The counsellor's presentation and demeanour should convey acceptance of patients as people.

1.2 Preserving Family Integrity

Genetic conditions may have a profound impact on the family unit, including both genetic relatives and relationships by marriage. Professionals should consider the integrity of the entire family, even if only one member comes for counselling.

2. Genetic Counselling

2.1 Counselling Competent Adults

Genetic counselling consists of (1) provision of all genetic and related information relevant to a family's needs; and (2) supportive counselling that enables a family or individual to make their own decisions after a process of gaining understanding of their own needs, values and expectations (Table 4). Optimum counselling can take place only in the context of available and affordable contraception and abortion for congenital disorders and available and affordable resources for caring for persons with disabilities. Adequate counselling does not mean simply providing information and leaving an individual or a family to their own devices. Adequate counselling means standing ready to help a family or individual work toward their own decisions about reproduction, testing, early diagnosis, prevention and treatment in a supportive and sympathetic environment.

Table 3
Reciprocal Responsibilities of Counsellors and Medical Geneticists

| Counsellors (competent adults) | Medical Geneticists |
|--|---|
| 1. Acquiring working knowledge of human genetics and its effects on people's lives; includes knowledge of the limitations of genetics and the importance of environmental and social contributions. | 1. Educating the public about effects of genetics on people's lives; includes education about the limitations of the genetic contribution and the importance of environment and society to health. |
| 2. Honesty in counsellor-medical geneticist relationship; includes full disclosure about medical status of relatives, existence of non-biological relationships (e.g., nonpaternity), and honesty about own reasons for seeking testing or prenatal diagnosis. | 2. Full disclosure of all test results that are relevant to health, including ambiguous test results, new and controversial interpretations, and differences among own colleagues in regard to test interpretation. |
| 3. Disclosure to relatives of genetic/medical information relevant to the relatives' health. | 3. Informing counsellors that it is the counsellor's ethical duty to tell relatives that the relatives may be at genetic risk. If the counsellor will not do so, the professional may inform relatives under certain conditions, when there is high risk of serious harm (if not forbidden by law). |
| 4. Disclosure of carrier status to spouse/partner if children are intended. | 4. Informing counsellors of ethical duty to disclose carrier status to spouse/partner if children are intended. |
| 5. If one's genetic status poses a danger to others, disclosure to the relevant authorities in the interests of protecting public health. | 5. Informing counsellors of their duties to disclose a genetic status that may pose a danger to public safety. If counsellor will not disclose and the risk is high, the geneticist should inform authorities. |
| 6. Awareness of potential misuses of genetic information by employers, insurers, schools, and other institutions, including possible unfair discrimination and stigmatization. | 6. Informing counsellors about possible misuses of genetic information by institutional third parties. |
| 7. Use of information provided in counselling to minimize or avoid harm to self or others. | 7. Unbiased presentation of information, insofar as possible. |
| 8. Timely presentation for testing, counselling, prenatal care, in order to avoid decisions about late abortion. | 8. Timely provision of indicated services or follow-up treatment (e.g., after newborn screening). |
| 9. Refraining from requests that serve no useful purpose (e.g., testing children for adult-onset disorders). | 9. Refraining from providing tests or procedures not medically indicated. |
| 10. Keeping the clinic informed of current address in order to allow recontact in case of new research findings. | 10. Keeping abreast of new developments. Recontacting counsellors on a timely basis regarding any developments relevant to their health or reproduction, unless otherwise instructed by counsellor. |
| 11. Expression of complaints or grievances. | 11. Ongoing evaluation and grievance procedures. |

Table 4

COUNSELLING GUIDELINES

1. Respect for persons and families.
2. Preservation of family integrity.
3. Full disclosure to counsellee of all information relevant to health, including ambiguous information. In prenatal or pediatric genetics, full disclosure means an unbiased description of family life with the affected person, as the person matures. Organizations for affected persons may provide supplementary information.
4. Protection of counsellee privacy from unjustified intrusions by employers, insurers, schools.
5. Duty to inform counsellees of relatives who might benefit from genetic information, or to directly contact relatives with genetic risk if not prohibited by law, if there is a high risk of serious harm to relatives in the absence of disclosure. Only medical information directly relevant to the relatives' own genetic status should be disclosed.
6. Accidental findings of nonpaternity should not be disclosed to a woman's partner.
7. Children and adolescents should be involved in decisions affecting them, whenever possible.
8. Nondirective approach, with some exceptions in cases of high risk of serious harm.

The counselling aspect of genetics services is best provided by a professional with a profound knowledge of genetics and who has had thorough training in counselling, whether medical doctor, nurse, social worker, or specially trained genetic counsellor.

If there is only one session, the counsellor should have documentation of the counsellee's test results in writing before the session, along with the complete medical record. It may be useful to have a preliminary session for blood sampling and planning of information collection as well as to prepare counsellees, in advance, about what they should expect from the main counselling session. Many people do not know why they have been referred for counselling or what will take place in counselling. The session will be more productive if people are told beforehand why they are going to counselling and what they can expect to gain from counselling.

2.1.1 Counselling Members of Different Cultural Groups

In counselling persons from different cultural or ethnic groups, an open mind and knowledge about the culture, religion, health beliefs, social values, and family structure of the counsellee's group are of paramount importance. The counsellor need not be from the same group as the counsellee, though in some instances this may be desirable. There are both advantages and disadvantages of having counsellors from the counsellee's own cultural or ethnic group. On the one hand, a counsellor from the same group may have first-hand knowledge of the values, beliefs, and practices prevalent in the counsellee's social

environment. On the other hand, even when counsellor and counsellee are from the same cultural group, there is often a considerable difference between them in education and social class. This difference may interfere with communication. Sometimes educated professionals are more inclined to be directive with less educated counsellees from their own communities than they are with counsellees from other communities.

Ideally, the counsellor should speak the counsellee's language. If an interpreter must be used, the interpreter should have special training in communicating with counsellees in medical settings, and should also have some basic knowledge about medical genetics.

2.1.2 Non-Directiveness

Counselling should be non-directive. Non-directiveness means that a counsellor must not direct the deliberations or decisions of a counsellee according to the views or values of the counsellor. This does not mean that the counsellor should be without a set of values. Most counsellees prefer to think that they are interacting with a morally concerned human being rather than a mere technician or provider of information. Non-directiveness means that the counsellor should be aware of his or her personal values and should not attempt to impose these personal values on counsellees, either overtly or covertly. The counsellor should not be an agent of a political or social entity or cultural group that seeks to impose its values on counsellees. Information should be presented in as unbiased a manner as possible. In counselling competent persons, geneticists should (a) help individuals/couples understand the present state of medical knowledge, their options, and the availability of social resources for people with disabilities, so that counsellees can make informed decisions; (b) suggest that while they will not make decisions for counsellees, they will support any they make; and, (c) tell counsellees that decisions, especially about reproduction, are theirs alone to make and refuse to make any for them (Fraser, 1974; Sorenson et al, 1981; Wertz and Fletcher, 1988).

If a counsellor holds strong opinions that he/she believes may lead to bias in counselling, it is better to be open with counsellees about these opinions at the outset, rather than presenting biased or selective information. Counsellors should be honest with themselves about their biases and should know their limitations. Counsellors who think that their opinions in a particular case may lead to biases in counselling should offer the counsellee a referral.

It may be appropriate to tell counsellees what other people in their situation have done, in order to illustrate a range of possible options. If referrals to other families who have experienced a similar situation are made, these families should represent the entire range of severity of the disorder in question and should also represent the range of opinions. Counsellors should be aware that families and organizations for people with genetic disorders can present biased views.

Some counsellees may ask what the counsellor might do if in their situation. If the counsellor will not reveal it, counsellees may try to guess the counsellor's opinion through verbal and non-verbal cues. Counsellors should be cautious in revealing such information. Telling a counsellee what the counsellor would do is not necessarily a directive approach, however, if carefully and sensitively presented. If the counsellor tells the counsellee what he or she would do in a particular situation, the counsellor must make it clear to the counsellee that the counsellor is not really in the counsellee's situation and cannot be in that situation, because the counsellor is a different person from the counsellee and has a different personal, family, social history, and situation. The counsellor should make clear that the

counsellor must make her/his own decision and that the counsellor's choice of action may be irrelevant to the counsellor's situation.

It is not appropriate to tell competent adults what to do in reproductive decision-making. Counsellors may be directive in health promotional activities that protect the health of adults, of fetuses (e.g., the maternal PKU diet) or the health of children. They should also refuse to perform or to offer referrals for non-medical services. Such refusals do not contravene the ideals of non-directive counselling.

2.1.3 Content of Counselling

Counselling should include a full description of the risks, diagnosis, symptoms, and treatment of the disorder(s) in question. For new parents, it is especially important to include all possible symptoms of the disorder in order to prepare them for the unexpected. If a counsellor is not expert on a particular disorder, the counsellor should refer counsellors to another team member, or to an outside expert. Counsellors should describe the development of a person with the disorder throughout the entire life course, and the effects of the disorder on family life. For disorders with a range of severity, the entire range of expression, from mildest to most severe, should be presented, together with an estimate of the likelihood of mild versus more severe outcomes. Use of visual materials should be encouraged. Movies or videotapes can convey the most information, by showing affected individuals and families in the course of their daily activities. Counsellors should not present people with genetic disorders in the impersonal, unclothed photographs that appear in medical textbooks.

Counselling should include information about financial costs, emotional costs, education, and both positive and negative effects on the marriage and family unit. Financial costs should include not only medical costs, but household costs (e.g., increased utility bills) that the family may reasonably expect. Counselling should also include information about available social and financial supports for persons with genetic conditions, assisted living (if applicable), and support groups. If "early intervention" (educational programmes in infancy or pre-school years) is available, the counsellor should direct the family to such programmes. Early intervention may be vital to later development. If a counsellor is not expert on the financial and social aspects of care, the counsellor should refer the counsellor to a social worker. Social workers should be integral members of all counselling teams. Counselling should include information about current research and should give a realistic assessment of future treatment possibilities.

2.1.4 Presentation of Risk

When risks are involved, whether these be genetic risks or risks arising from a procedure or treatment, counsellors should present these risks in several different forms: proportional (1 in 4, etc.), percent (25%), and in a verbal form (e.g., higher than the average for the general population). Counsellors should be aware that many people have difficulty interpreting risks. There is a tendency for counsellors to regard a proportional risk as higher than the same risk given as a percent. Hence the importance of presenting a risk both ways. Some people may tend to overestimate small risks or to underestimate high risks (as long as these do not reach 100%). Counsellors tend to interpret the same numeric risks as lower than do counsellors (Wertz et al, 1986). Counsellors also tend to interpret risks in binary ("either/or") form, whatever the level of risk. Counsellors should not expect counsellors to make decisions primarily on the basis of a risk figure.

The difficulty that some people experience with risk interpretation underscores the need

to provide the fullest information possible about the disorder in question. In helping counselees to make decisions, counsellors should ask counselees to envisage the future consequences of each alternative, considered over the life course. Counselees should be asked to consider the consequences of each choice for themselves, their spouse or partner, family, and community.

2.1.5 Contexts of Counselling: Settings and Scheduling

Persons receiving unfavourable diagnostic test results (prenatal, pediatric, presymptomatic, or adult) should always receive full counselling. This should extend over several sessions if necessary. Adequate time should be allotted at each session for counselees to be able to present their concerns in full and to receive supportive counselling. Often this may require 45-60 minutes or more. No arbitrary time limits should be set, however, as individuals vary considerably. In the interests of efficiency, most counselling will take place in centralized settings such as clinics, hospitals, or community health centres to which counselees travel. These centres should be available to public transportation. Appointments should be scheduled with consideration for the counsellee's work schedule and also the schedule of public transportation. The waiting time before scheduling the initial appointment, which should be used to collect family information, should be as brief as possible, and counselees should be served promptly on arrival at the clinic. If a waiting period is usually necessary after arrival at the clinic, counselees should be warned of this in advance.

Sometimes alternative settings may be optimal in enhancing communication. Some people feel more comfortable talking with the counsellor in the familiar setting of their family physician's office than in an unfamiliar clinic. In some rural areas it may be appropriate to provide some basic types of counselling (such as pre-screening information) during home visits by midwives or community health workers.

Privacy is essential to good counselling. Counsellor and counsellee must be able to meet in a private room with the door closed.

Child care should be available, at no cost, for individuals or couples who bring children with them to counselling, so that the individual or couple can talk with the counsellor without interruption. Usually it will suffice if there is someone available to take the child(ren) out of the room.

All persons should receive some form of basic counselling before screening, diagnostic testing, or prenatal diagnosis. In some cases, this counselling may provide information only, through printed or audio materials, movies, or videotapes. Such information should be standardized throughout a health care system to make certain that all receive equal information. Verbal information should not be the sole source of information. When printed, audio, or visual materials are used to provide pre-test information about the test(s) and the disorders(s) in question, counselees should also have the opportunity to discuss the test(s) with a knowledgeable person (perhaps a community health worker) before testing.

2.1.6 Non-Discriminatory Language

Choice of language can have a powerful effect upon one's perceptions of people with genetic conditions. Counsellors should describe individuals with genetic disorders as persons first, rather than defining them in terms of their conditions. The phrase "child with cystic fibrosis" describes a child who happens to have cystic fibrosis, whereas the phrases "cystic fibrosis case" and "cystic fibrosis child" present the disease as the foremost consideration and

the child as secondary. The usage of "person with [name of disorder]" is always preferable to "[name of disorder] patient." "People with disabilities" is preferable to "disabled people" or "the disabled." "Disorder" or "condition" are preferable to "disease," because some genetic conditions are not diseases. The terms "burden" and "suffering" should be used carefully; many genetic conditions are not burdensome and do not cause suffering to those who have such conditions. A condition, may, however, cause burden or suffering to a family (or community), even with social supports. If such terms are used, the favourable as well as unfavourable aspects of a condition should be presented. The terms "positive" and "negative" should not be used in presenting test results to counselees, because most counselees will find these terms confusing. The term "family history" is preferable to terms employing the word "pedigree", which some people associate with animals only.

2.1.7 Counselling Spouses/Partners

If a couple intends to have children, both partners should be counselled together. The counsellor should encourage each partner to express her/his views on family life with a child with a genetic disorder, in the presence of the other partner. Frequently partners hold different views about various aspects of caring for a potential child with a disorder (Sorenson and Wertz, 1986). It is important that these views be aired and discussed, preferably before a child is conceived.

2.1.8 Referrals to Organizations for Persons with Genetic Conditions

Information about lay organizations may in some countries and situations be an appropriate part of counselling for those with pathological findings, including that from prenatal diagnoses. Counsellor follow-ups should be voluntary, however.

2.1.9 Summary of Counselling Session Provided to Counselees

At the end of each counselling session, the counsellor should summarize the contents of the session briefly from the counsellor's point of view. The counsellor should then ask the counsellee to summarize the session briefly from his or her point of view. The purposes of this final summarizing are (1) to refresh the counsellee's memory; (2) to help the counsellor evaluate the counsellee's level of understanding of medical/genetic knowledge; and, (3) to help the counsellor evaluate the counsellee's need for further supportive counselling or referrals.

The counsellor may record these summaries, either in writing, or (with the counsellee's permission) on tape. The counsellee could receive a written copy of the summaries by mail after the session (but only if the counsellee gives permission and if written information can be kept confidential) and/or a tape of the summaries if the counsellee has access to a tape recorder. The purposes of providing counselees with a tangible record summarizing the session are (1) to aid the counsellee in retaining complex information; and, (2) to provide information to other family members not present at the session, if the counsellee so wishes. The summaries should also become part of the counsellee's medical record kept on file by the counselling centre.

2.1.10 Materials Provided to Counselees

Counsellors should provide educational materials appropriate to the counsellee's level of literacy. If a counsellee cannot read, tape-recorded, pictorial, or video-taped materials may be appropriate. In some cases, use of these may have to be on-site at the centre. At

the other end of the spectrum, many educated counselees will desire information beyond that usually presented in informational brochures. Some counselees will wish a list of publications available at their libraries, and some will wish to look at the medical literature itself. Counsellors should be able to provide up-to-date lists of publications at all levels.

2.1.11 Evaluation of Counselling

Those providing counselling should have evaluation measures in place to assess the quality of communication, counsellee understanding of information, and usefulness of counselling to counsellee decision-making. Evaluations should be reviewed on a regular basis, with the aim of improving communication. Counselling should not be evaluated in terms of numbers of tests, prenatal diagnoses, or abortions subsequently performed. Using numbers of procedures as a measure of effectiveness in counselling may lead to directiveness on the part of counsellors, who may urge their counselees to be tested. Numbers of births (of children with genetic conditions) averted should not be used as a measure of effectiveness of counselling, although public health authorities should keep such data for epidemiological purposes. Effectiveness should be judged only in terms of (1) successful communication of information, as evidenced by counselees' understanding; and, (2) counselees' reports (or other evidence) that counselling assisted counselees to make decisions that were best for the counselees, in the light of their own values and family goals.

Evaluation should include (1) record review; (2) peer review, with peers attending each other's counselling sessions on a regular basis (with the counsellee's permission) and criticising each other's work in a non-judgmental manner; and, (3) counsellee review, using periodic surveys or interviews.

2.2 Counselling Children and Adolescents

2.2.1 Involving Children in Decisions

Whenever possible, children and adolescents should be involved in decisions about their own treatment. Children over the age of 7 may understand some basic aspects of disease and treatment affecting them. Their verbal assent should be sought, but should not be binding. Parents should make decisions regarding therapy or preventive measures. (For further discussion, see 9. Testing Children below.)

As a child enters adolescence, the child's wishes should carry greater weight. There is no precise age at which a child or adolescent's wishes should be considered equally with those of the parents. This will vary on a cultural, family, legal, and individual basis. The maturity of a child or adolescent to contribute to a decision, to initiate, continue, or discontinue testing or treatment should be assessed on a case-by-case basis, using generally accepted criteria for competence.

2.2.2 Requirements of Competence

Knowledge of fact alone does not constitute competence to request or consent to testing. Competence includes (1) voluntariness; (2) "reasonable outcome" of a choice in terms of the individual's and family's social and cultural situation, values, and life style; (3) "rational" reasons for the choice that would be understandable to most reasonable persons; and, (4) understanding of risks, benefits, and alternatives, including knowledge of both facts and implications (Katz 1972, 1984). Piaget (1965) suggested that the type of formal operational thought necessary for competence began at about 11 and was well developed at

14. There may be difficulty in judging whether a minor's request or consent is truly voluntary, however. Caution must be exerted.

2.3 Counselling Persons with Diminished Mental Capacity

Non-directive counselling (refraining from direct advice to protect and enhance the autonomous choices of counselees) is a commitment of genetics professionals. This assumes that all relevant facts are known to the counsellee, and efforts are made to encourage the counsellee to consider the facts in the context of his or her beliefs and values.

A possible exception to non-directive counselling can arise in genetic counselling with incapacitated patients, especially when genetic harm to others is a potential danger. These patients may be mentally ill, severely retarded, or abusers of alcohol or drugs. Some patients may be severely disadvantaged in communication because of poor education, although they are of normal intelligence. For these reasons such patients may be functionally unable to weigh the significance of genetic risks.

The actual incidence of this type of situation and geneticists' response to it needs careful study. In principle, giving direct advice to relatives of incapacitated counselees or to impaired counselees themselves is ethically acceptable, in exceptional cases, if the likelihood of harm to others is great and if the geneticist has informed the patient or relatives in advance of counselling that directive counselling may be indicated.

When persons of diminished capacity desire to have children, the counsellor must weigh: (1) their understanding of any risk to themselves and the child; (2) their capacity to rear the child; and, (3) social supports. One example would be a woman with fragile-X syndrome who desires to have children. She is mildly retarded and does not understand the increased risk to her offspring, despite repeated efforts at counselling. Directive counselling, with involvement of the family, could be the ethical approach, in some cases.

2.4 Competent Adults who Abdicate Moral Autonomy

In the rare event that a competent adult refuses to participate in the non-directive model of genetic counselling and insists that the professional make the decisions, all decisions should be in the best interests of the individual. In these rare instances the professional-patient relationship follows the fiduciary model, wherein a client voluntarily assigns the power of decision to an expert. This model should be used only as a last resort and only if the counsellee insists on it.

2.5 Effects of Professionals' Gender

Gender differences in counselling suggest that counselees should ideally be offered the opportunity to meet with counsellors of both genders in order to cancel out possible gender biases (Wertz, 1994).

3. Informed Consent

Screening (with the exception of mandatory newborn screening), diagnostic genetic testing, prenatal diagnosis, treatment, and research should be preceded by informed consent. Informed consent means that the counsellee understands the risks, discomforts, and benefits of the procedure(s) to be performed and is aware of the various alternatives, including the

alternative of not performing the procedure. Informed consent means that the counsellor consents voluntarily. The elements of informed consent appear in Table 5 overleaf (Fletcher et al, 1983). The purpose of informed consent is to make certain that counsellees understand possible effects of procedures and that they are willing to undergo these procedures.

Formal informed consent, in the form of a written document, is not necessary for procedures that constitute part of routine care. Formal informed consent should be required, however, for experimental procedures or risky procedures, if the person is competent to consent. All persons having genetic screening or testing, however, including the parents of newborns, should be informed before testing about the major characteristics of the disorder(s) screened or tested for, the limitations of the test (possible false positives, false negatives, or indeterminate findings), the risk of receiving unfavourable test results, and possible consequences of such a result. Possible socio-economic consequences of an unfavourable test result, such as loss of health or life insurance, refusal of employment, discrimination by schools, adoption agencies, etc., should, where applicable, be included under the description of risks. If a test may reveal nonpaternity as an incidental finding, this also should be included in the description of risks. If results may be ambiguous, counsellees should be informed of this possibility. Women receiving biochemical screening during pregnancy should be informed, before screening, that there is a chance that they could ultimately face a decision about abortion. All counsellees should be informed of their rights to refuse screening or testing (except for mandatory newborn screening.)

Information should be presented simply, in non-medical terms, and in the counsellor's own language. It is not sufficient to provide information in the form that an ideal "reasonable person" could understand. Individuals and families, especially in multicultural societies, have different means of understanding and assimilating information. Informed consent, whether informal (verbal) or formal (written) is only valid if it represents true understanding.

Genetics professionals should attempt to evaluate understanding, especially for procedures involving higher risks. One way to evaluate counsellees' understanding is to ask them to describe the procedure, its purpose, and its risks/discomforts in their own words. If the professional is not satisfied with the response, the professional should go over the information again. If a counsellor cannot understand the information, despite the best efforts of the professional and other educators, and if the procedure is not experimental, the professional may proceed if in his or her judgment the procedure provides significant benefit, and if the counsellor wishes to go ahead. To withhold a non-experimental procedure because a competent counsellor cannot understand it, despite the professional's repeated efforts, is judgmental.

In the case of competent adults, no person should be permitted to give consent for another. Although decisions about screening, testing, and prenatal diagnosis may be family decisions, consent should be on an individual basis.

Whenever possible, children and adolescents should give assent for testing and treatment. (See Part 1, 4.5 above).

Table 5

ELEMENTS OF INFORMED CONSENT AND ASSENT FOR RESEARCH

(*relevant to genetic testing)

Elements of Informed Consent

1. State and describe
 - *a. Research nature of study (if experimental)
 - *b. Purpose of study [or test]
 - c. Duration of participation
 - *d. Procedures to be followed
 - *e. Which procedures are experimental
2. Describe
 - *f. Reasonably foreseeable risks
 - *g. Discomforts
3. Describe
 - *h. Benefits to the subject
 - *i. Benefits to own group (peers)
 - *j. Benefits to others
4. Disclose
 - *k. Alternative procedures or treatments
5. Describe
 - *l. Confidentiality of records identifying the subject
6. Explain, if the project involves more than minimal risk
 - m. Policy on compensation for injuries due to research
 - n. Availability of medical treatment for such injuries
 - o. Source of further information
7. Explain
 - p. Whom to contact for questions about research, or
 - q. In event of research injury
8. State
 - *r. Participation is voluntary
 - *s. No loss of benefits on withdrawal
 - *t. May withdraw at any time

Elements of Assent for children over age 7

- *1. Purpose of study
2. Duration
- *3. Procedures
4. Role of child
5. Knowledge of research participation
- *6. Risks/side-effects
- *7. Benefits to self
- *8. Benefits to peer group
- *9. Benefits to others
- *10. Alternative treatments [or alternatives to testing]
- *11. Confidentiality
- *12. Freedom to ask questions
- *13. Voluntary participation
- *14. Freedom to withdraw
- *15. Right not to know results

4. Rights to Referral

If a physician is unable or unwilling to perform a medically indicated service for personal moral reasons, the physician is obligated to refer the counsellee to someone who will perform the service, provided that (1) the service is legal, and (2) the service is a medical service related to the diagnosis, prevention or treatment of disease. A physician should not refer for sex selection in the absence of X-linked disorder, because sex is not a disease.

Sometimes laws are created by dominant political, cultural, and religious interests and may not necessarily be fair to all persons in a society. In nations where abortion is forbidden for most purposes, it may be impossible to obtain a legal abortion after prenatal diagnosis (Penchaszadeh, 1993b). As in other areas of medicine, the physician has a moral duty not to abandon counsellees after a diagnosis. To do so would be a breach of the physician-patient relationship. Although a full range of genetics services, including prenatal diagnosis, should be available in every nation, individual physicians may choose not to perform prenatal diagnosis for reasons of conscience, if they oppose abortion. A physician who performs prenatal diagnosis should respect women's choices and help women to find safe, affordable medical care so that they can act on these choices. In nations where abortion is illegal, a physician who performs prenatal diagnosis owes the counsellee a referral for a safe, affordable abortion in another country.

In general, physicians and other professionals owe counsellees a referral whenever the professional believes that his or her own personal beliefs may compromise communication or patient care.

5. Duty to Recontact

Genetics professionals have an ongoing duty, unless instructed otherwise by the counsellees, to inform counsellees and families about new tests and treatments. In genetics, the ethical responsibility to follow up and recontact families may extend for several generations. The physician's ethical duty extends beyond those individuals who have presented themselves as counsellees. Ideally, all family members at genetic risk should be informed of all new developments, provided that it is possible to find them and that they are willing to be informed.

In practice, this ideal may be impossible to carry out. At a minimum, genetics services providers should urge counsellees and relatives to contact the clinic regularly about the possibility of new developments and/or to provide the clinic with updated addresses so that the clinic can contact them.

The need to recontact indicates the significant benefits of genetic registers (Berg, 1983). As noted in section 7.2.8 below, such registers would cause harm to individuals and families only if data protection is not strict. Mechanisms for ensuring privacy must be established.

6. Screening and Testing

6.1 Definitions and Requirements for Programmes

6.1.1 Screening

Screening is applied to large-scale populations with no known excess risk to individual persons (see also Bankowski and Capron, 1991; Council of Europe, 1992; Nuffield Council, 1993). Screening is frequently part of government-sponsored public health programmes (Science Council of Canada, 1990). Screening may be a preliminary procedure that identifies persons at elevated risk but does not provide a definitive diagnosis. Biochemical screening, such as maternal serum alpha-fetoprotein measurements in pregnancy, is an example. It identifies fetuses at elevated risk for Down syndrome or neural tube defects, but does not result in diagnosis unless followed by amniocentesis or chorionic villus sampling, which are then diagnostic tests rather than screens.

Screening may also be used to identify persons with higher-than-average susceptibilities to common diseases such as heart disease. Sometimes screening results in a definitive result, as in newborn screening for PKU or carrier screening for haemoglobinopathies. Screening programmes require that treatment or preventive measures are available for a disorder and that treatment or prevention is likely to make a difference to the individual's health. Nations instituting screening programmes must provide timely and affordable treatment or prevention of the disorders screened.

Some public health programmes screen healthy people for carrier status. In the past, screening programmes undertaken without the knowledge or cooperation of populations to be screened have failed, sometimes after accusations of ethnic discrimination against targeted groups (United States, 1973). All screening programmes must be preceded by education of the populations or communities to be screened. If a particular ethnic group is to be targeted because of elevated risk for a particular disorder, screening should be undertaken with the active cooperation of leaders and members of this group.

Proven measures of prevention or treatment must make a substantial difference for at-risk persons or families identified. The meaning of "substantial difference" will vary in different nations, according to the public health resources available. Sometimes commercial interests attempt to create markets for screening and treatment, without the knowledge, cooperation, or interest of the community (e.g., for Gaucher disease in the USA). Such attempts should be resisted.

6.1.2 Diagnostic Testing Compared with Screening

Diagnostic testing differs from screening in regard to the population served (Berg, 1991). Whereas screening applies to populations with unknown risks to individuals, diagnostic testing is offered to individuals and families who are at higher-than-average risk because of family history of a genetic disorder, history of environmental exposure, advanced maternal age, or positive results of a prior screening procedure, or clinical signs in the persons to be tested. Diagnostic testing, unlike some screening, has as its goal definitive diagnosis.

Screening is sometimes part of routine medical care and, if it provides only information concerning risk levels without definitive diagnosis, is sometimes carried out without informed consent, although requiring informed consent is the most ethical course of action.

6.2 Voluntary *versus* Mandatory Screening

Screening should be voluntary and should be preceded by informed consent and information/counselling, with one exception: screening of newborns, if and only if, early diagnosis and treatment would benefit the newborn.

6.3 Newborn Screening

Societies have an ethical obligation to protect their most vulnerable members, especially if these people cannot protect themselves. Newborns deserve the special protection afforded by mandatory screening for disorders where early diagnosis and treatment favourably affect outcome. In arguing for inclusion of a disorder on the list of mandatory screens, public health authorities should be able to prove that early diagnosis and medical treatment make a difference for the population of newborns with the disorder. The psychosocial benefits of simply having a diagnosis, in the absence of treatment, are not sufficient to justify mandatory screening. For example, screening for fragile X syndrome is not warranted because there is no evidence of medical benefit to the newborn. To justify mandatory screening, benefits must accrue to the newborn. Screening for dyslexia (if this became possible) would not be warranted unless benefits occurred in infancy. Such screening would be better undertaken on a voluntary basis later in childhood. Screening should not be mandatory if its primary purpose is to identify and counsel parents who are carriers before their next pregnancy (e.g., for Duchenne muscular dystrophy). Parental carriers are best identified through public education about potential risks for various disorders, followed by voluntary testing on an individual basis, preferably before conception.

Newborn screening should be conducted within the optimum time frame for early detection and treatment. If the maximum sensitivity of a test occurs at some point after birth and possibly after early discharge from the hospital, it is imperative to follow up and test the newborn at this time. A just health care system should provide outreach to all newborns, free of charge, at the time when screening is most likely to detect a genetic disorder and before a genetic disorder, if present, can cause permanent damage to the newborn. Follow-up visits have proven feasible in several nations.

Centralized hospitals are optimal, but cannot be depended upon as efficient or appropriate avenues of screening if many newborns are discharged within 24 hours after birth or many births take place at home. Home visits by community health workers or nurses several days after birth are fair and appropriate avenues of providing newborn screening at the optimum time.

The primary purpose of mandatory newborn screening is to benefit the newborn through early treatment. Some treatments (e.g., for maple syrup urine disease or PKU) must be instituted immediately in order to be effective. It makes no sense to provide screening if timely treatment is not available. Nations instituting newborn screening programmes are ethically obligated to provide available, affordable, and timely treatment for each disorder in a screening programme. If a nation is unable to provide affordable and timely treatment to all for a disorder, that disorder should not be included in mandatory newborn screening.

If multiplex screens are used, the multiplex should be limited to only those disorders included in the mandatory screening programme. If additional tests are done at parents' request, these should be in a separate analysis.

Newborn screening for treatable disorders may reveal carrier status in the family. When this occurs, the parents or family members should be informed if this has health consequences for the newborn or family. The parents may choose whether to be tested to identify carrier-carrier couples. In informing parents of the newborn's status, professionals should be careful to prevent parental misconceptions that may stigmatize the newborn. The purpose of informing parents about the newborn's carrier status is for the benefit of the parents' own reproductive plans and the avoidance of harm to their future children. Geneticists should weigh these potential benefits against potential harms to the newborn in each case, and should disclose only if benefits outweigh harms.

Information to parents should precede all screening and diagnostic testing, whether voluntary or mandatory. Unfavourable test results should be followed by full genetic counselling.

Test results, including information from blood spots used in mandatory screening, should become part of the child's medical record and should receive the same protection of confidentiality as applied to medical records.

6.4 Screening in the Workplace

Screening in the workplace for genetic susceptibility to occupationally-related diseases is forbidden in some countries. Nevertheless, screening may be in the worker's best interest, if a nation's laws adequately protect the worker's rights to employment, medical care, and economic support. Screening may offer protection for some workers in nations where workplace safety is inadequately regulated. Screening in the workplace should not be used as a substitute for making the workplace safer. All screening, whether before or after hiring, should be voluntary, and workers should be informed of their own test results and the meaning of these results. Refusal to be screened should not prejudice hiring or continuation of employment. Employers should not have access to test results even with a worker's consent. If a test result indicates that a worker is at high risk, and if the workplace cannot be made safer for susceptible workers, the worker should be transferred to a safer job within the company, at the same pay. If transfer is not possible, the worker should be given the choice of whether to stay in his/her former job or whether to leave the company's employment, after full counselling about the consequences of each alternative.

Genetic monitoring is regular periodic examination of all workers for chromosomal breakage or other evidence of genetic damage from exposure. Unions often favour monitoring over susceptibility screening, because (1) it takes place after workers have already been hired, and (2) it may be more likely to lead to beneficial changes in the workplace than susceptibility screening (Draper, 1991). The drawback is that it reveals damage that has already taken place, rather than preventing such damage. A combination of screening and monitoring, on a voluntary basis, with all results disclosed to the worker in a timely fashion, and with full protection of employment, is probably the most ethical approach (Berg, 1982).

6.5 Jobs Involving Public Safety

Sometimes a worker's genetic disorder may affect public safety. This is most likely to occur in the incipient stages of a late-onset disorder, before diagnosis (e.g., the air traffic controller who cannot follow the video monitor adequately because he/she is in the early stages of Huntington's chorea). Ideally, regular physical examinations of all employees in jobs involving public safety would identify persons who pose a risk to others. Unfortunately, this is not always possible, whether because of the characteristics of a particular disorder, the

length of time between routine examinations, or the comprehensiveness of the examinations. In cases where an employee has a family history or otherwise elevated risk for a disorder that may pose a risk to others, an employer may require testing as a condition of continued employment. Persons holding public safety jobs include all those who could seriously endanger members of the general public while carrying out their work. They include those who operate motor vehicles on the job, police and fire fighters, physicians, all persons whose jobs involve carrying a weapon, and persons responsible for national defense policy.

Persons with unfavourable test results should be allowed to continue in a job as long as physicians determine that their present status poses no significant risk to others. If and when they pose a risk, they should be placed on disability or retirement benefits. A worker's genetic status should not be used to force early retirement. A full review process should be in place to protect workers from discrimination. The worker should continue to receive salary or wages while under review, but should be placed on leave from the job.

6.6 Premarital Screening for Carrier Status

Premarital screening for carrier status for disorders common in a community should be encouraged. Premarital screening allows couples a fuller range of options than post-marital screening. All such screening, however, should be voluntary, with the cooperation of the community, and preceded by full education. Premarital screening should not be required by law, as this violates personal autonomy.

7. Disclosure and Confidentiality of Test Results

The most frequently occurring ethical dilemmas in clinical practice involve disclosure. Sometimes geneticists fear that disclosure of psychologically sensitive information to a counsellee will lead to more harm than benefit. Confidentiality means an agreement not to reveal information. The agreement may be explicit or may be implicit in the physician's role. This duty is universally respected. Privacy, a largely Western concept, means, in addition to the right to be left alone and free from unwarranted intrusion, also ownership of one's self, including the body and all things pertaining to it, including medical information. Privacy is part of the principle of autonomy or respect for persons.

7.1 Preparing Counsellees before Testing

In general, it is best to prepare counsellees and families for possible disclosure dilemmas before they undergo testing. This approach minimizes psychological shock and hasty decisions. Pre-test counselling should include the information that in some cases test results may be ambiguous or conflicting. If a test conducted for another purpose may incidentally reveal non-paternity, unacknowledged adoption, or other non-biological relationships, couples should be warned of this possibility before testing. The mother should be counselled individually before a couple is seen together, so that she can withdraw from testing if nonpaternity is a possibility, without revealing to her partner the reason for her withdrawal.

If a test will reveal which parent carries the genetic material that has caused a disorder in a child, the mother should be forewarned, because the woman is often blamed for a child's condition. She may decide to withdraw from testing. If she decides to go ahead with testing, both parents should then receive adequate pre-test education and counselling to prevent marital strife about possible consequences of genetic testing. If both agree, both should

receive the information, provided that counselling has established that disclosure to the non-carrier will not harm the carrier.

Counselees should also be informed, before testing, about any employers, insurers, other institutional third parties, government agencies, or others who in many countries may lawfully seek access to or be able to require access to their test results. Counselees should be informed in advance of the clinic's policy on disclosure to relatives at genetic risk (see 7.2.2 below), and relevant laws or regulations.

7.2 Situations Involving Disclosure and Confidentiality

7.2.1 Full Disclosure of Clinically Relevant Information to Counselees

Full information is a prerequisite for free choice. Professionals should disclose all test results relevant to an individual's own health or the health of a fetus, including results indicative of any genetic condition, even if the professional regards the condition as not serious. Those who will bear and rear the child should decide, after receiving full and unbiased information, about the effects of the condition on their family, and its social and cultural situation. Test results should be disclosed even if ambiguous or conflicting. New or controversial interpretations of test results should also be disclosed. Although some disclosures (e.g., of ambiguous prenatal test results) may cause anxiety or distress, disclosure is preferable to concealment, because disclosure shows respect for the counsellee as a person and allows the counsellee to make decisions.

Although counselees may have a "right not to know" genetic information, the right not to know presumes that counselees understand what it is that they have chosen not to know about. The complexity of genetic information, especially from multiplex tests, makes selective disclosure of medical/genetic information difficult, and this alternative should not be encouraged. (See 7.2.1e below, "Selective non-disclosure".) In the future, as people learn more about genetics, they will be better prepared for troubling disclosures.

Full disclosure is necessary to the open communication and trust that should mark the physician-patient relationship. If vital facts are edited out of the communication by the physician, the relationship is less than optimal and can be harmed. If the counsellee later discovers non-disclosure, confidence in physicians could well be shaken or undermined and result in further harm. Counselees should be informed in a timely and convenient manner. The informer should be a health professional in person, but timeliness is essential. Therefore in some cases a telephone call or a home visit by a rural health worker may be acceptable.

Disclosure of Psychologically Sensitive Information

In situations where the nature of the information to be conveyed (e.g., XY genotype in a female) could cause grave psychological harm to a counsellee or family, the "therapeutic privilege" of delayed disclosure is allowable. Situations justifying delayed disclosure include immaturity (chronological or psychological) and lack of education. The therapeutic privilege presumes full disclosure, but postpones it until the counsellee is psychologically and cognitively ready. The therapeutic privilege is sometimes overused or is used to justify medical directiveness. It is wise, where possible, to obtain a second opinion about the probability of psychological harm before making a decision to defer or delay disclosure. Therefore the therapeutic privilege should be invoked only after consultation with a mental health professional knowledgeable about genetic disorders and their psychological consequences. The professional should determine before disclosure whether psychological

help will be available. In the absence of such help, and when assessment shows that emotional harm is possible, nondisclosure or delayed disclosure of the full scientific facts may be justified.

Disclosure of Normal Test Results

Counselees and families frequently worry when a test is taken and no results are communicated. Normal test results are of great interest and importance to individuals and families. All normal results should be communicated in a timely fashion.

Non-Medical Results

Test results without direct relevance to health (e.g., nonpaternity, fetal sex in the absence of X-linked disorders) may be withheld if this appears necessary to protect a vulnerable party. Counselees also have a right not to know this information if they so choose, and they should be informed of this right before testing. In some countries, handling of non-medical results is regulated by national law.

Prior Disclosure to Another Party

Sometimes a counsellee asks that test results be disclosed first to someone else. Usually the person to receive the disclosure is a spouse or family member. The request may be honoured, but only after careful counselling of both parties to make sure that the request is voluntary. The professional has an obligation, however, to make sure that the results reach the counsellee her/himself in a timely fashion.

Selective Non-disclosure at Counsellee's Request

Counsellees may have a "right not to know" genetic information if they do not wish to know. Usually they exercise this right by deciding not to be tested. Sometimes, however, a counsellee wishes to have a test but to be told only some types of results (e.g., the woman who has prenatal diagnosis for Down syndrome but does not wish to be told if there is a sex chromosome abnormality). Such choices may be honoured, provided that the counsellee understands the possible consequences of selective knowledge. In view of the number of genetic conditions that a test may disclose, however, providing medical/genetic information selectively is usually not in the counsellee's best interests and should not be encouraged. Counselees have a right not to know non-medical information such as false paternity or fetal sex. Agreements about disclosure or non-disclosure of such information should precede testing.

7.2.2 Confidentiality when other Family Members are at High Risk

In genetics, the true patient is a family with a shared genetic heritage (Berg 1989). Family members have a moral obligation to share genetic information with each other (Berg, 1994). The ethics of disclosure of genetic risks begins with intra-familial duties to warn and protect family members from harm, and these duties are not confined to the immediate family. Identified patients or parents of an affected child (Andrews, 1987) have an ethical duty to inform relatives in the extended family, once they are informed themselves about the condition. This duty arises from kinship bonds and the ethical principle of non-maleficence. A basic function of the family itself is protection from harm for its members. However, those at risk must first learn about their risks. Physicians, especially medical geneticists, are the primary mediators of genetic knowledge in society today. Medical geneticists are entitled

to ask assertively, if not to require, that the identified patient or parents help in contacting relatives so that they may be informed about specific risks. The first contact with the counsellee or key family members ought to include discussion about family involvement and responsibilities to disclose findings. Also, depending upon the degree and magnitude of harm that may occur from non-disclosure, the counsellor should discuss the limits of confidentiality at the outset.

It is the counsellee's moral obligation to tell relatives at risk about a diagnosis and/or results of presymptomatic tests, so that these relatives can choose whether to be tested themselves. It is also a counsellee's moral obligation to provide blood, saliva samples or other specimens, so that relatives can have genetic tests. It is the medical geneticist's moral obligation to remind counsellees of these obligations. "Nondirective counselling" is not appropriate in these situations. Usually counsellees will cooperate if repeatedly urged to do so.

Sometimes a counsellee prefers that the professional, rather than the counsellee, tell relatives. The counsellee may feel embarrassed about transmitting bad news, but also thinks that the relatives should know. The professional should offer counsellees the option of having the professional tell the relatives, at the counsellee's request. If the relatives live at a distance, the geneticist should offer referrals to professionals living near the relatives who can tell them, again at the counsellee's request.

In asking counsellees to tell their relatives (or to have the professional do so), the professional should keep in mind the relatives' rights to confidentiality as well as the counsellee's rights to confidentiality. In some cases, as when a family feud exists, the counsellee may use implicit information about the relatives to harm the relatives' opportunities for marriage or employment. The professional should guard specific information about the relatives' potential risks.

Genetic information is both uniquely individual and the shared property of families. Laws affecting confidentiality, privacy, and rights to information have in general not yet taken account of this unusual medical situation. What a physician may legally do with genetic information at this point in time will vary among nations. In the future, laws should be revised to reflect the shared nature of genetic information while protecting the privacy of individuals. Meanwhile, medical geneticists should keep in mind two well-known duties in medicine, both of which may be supported by laws in many countries. They are (1) the duty to maintain patient confidentiality; and (2) the duty to warn third parties, such as relatives at genetic risk, of harm.

If possible, the medical geneticist should attempt to warn relatives who are at high risk of serious harm, even against the counsellee's wishes, provided that the four conditions described below are met. The geneticist should also keep in mind the relatives' moral right not to know their own genotype and not to have diagnostic testing, provided that the exertion of this wish does not cause harm to others. The warning about genetic risk should take the form of a general announcement informing relatives that they may be at elevated genetic risk and inviting them to seek consultation with a geneticist if they wish. A general warning falls under the heading of public health information to persons at elevated risk and does not infringe on rights not to know genetic status. The relatives may choose not to seek counselling and may thus exercise their rights not to know. The geneticist should take care not to identify or describe the genetic status of the original counsellee, except with the counsellee's permission.

In rare cases an individual refuses to disclose or to permit disclosure of information to relatives at genetic risk. The genetics professional may, unless it is prohibited by law, override individual confidentiality if the following four conditions are met (United States, 1983):

- All efforts to persuade the individual to disclose the information voluntarily have failed.
- There is a high probability of harm to the relatives (including future children) if the information is not disclosed, and there is evidence that the information could be used to prevent harm.
- The harm averted would be serious.
- Only genetic information directly relevant to the relatives' own medical status would be revealed. Information relevant to the counsellee must remain confidential.

Persons who may have a need to be informed should include the siblings or children of persons with autosomal dominant disorders, with X-linked disorders, or with disorders for which the mutation may undergo expansion in succeeding generations (e.g., myotonic dystrophy, fragile X syndrome). These persons are at high risk. In cases of autosomal recessive disorders or carrier status for such disorders, the risks to relatives are often small, because the chance of a relative marrying another carrier is slight. Overriding confidentiality is not justified for autosomal recessive disorders or sporadic conditions.

Professionals should be legally permitted to disclose information if all of the four conditions listed above are fulfilled; in other words, there should be no legal penalty for disclosure. Professionals should be legally required to disclose information about relatives' own genetic risks if the relatives ask. Professionals should not be legally required to disclose such information to relatives who do not ask. Although in many cases relatives may not be aware that they are at risk and therefore do not ask, finding these relatives and disclosing information to them places too much extra burden on the doctor. If relatives can be found easily, however, disclosure could be the most ethical course of action.

Overriding confidentiality may have a legal as well as ethical basis. Genetic information is not the sole property of individuals, but is shared among family members. In the future, ownership of information laws should be revised in order to reflect the dual nature of genetic information: individual and familial. It would be unethical to conceal genetic information from its owners, who include blood relatives with genes for a disorder.

It is also unethical to reveal information pertaining to one individual to others, without the individual's consent. It could be ethical, however, to locate and disclose to family members the fact that they may be at genetic risk and to ask them to come to a clinic for testing, through the counsellee, if at all possible. The professional need not, and should not, tell the family members the source of the information. The professional thereby fulfils the duty to warn third parties of harm without disclosing the name or diagnosis of the counsellee. Family members may, of course, learn the counsellee's diagnosis indirectly as a result of their own counselling and tests. This is probably impossible to prevent. Nevertheless, the counsellee should not be able to prevent others from learning genetic information about themselves.

Cases of outright counsellee refusal to contact relatives will, however, continue to occur. If geneticists have informed counsellees at the outset about the need and duty to inform other family members who have a reproductive or health risk, and have also informed counsellees that confidentiality is limited by this moral duty, medical geneticists have laid the groundwork for action if the counsellee subsequently refuses to contact relatives, unless action by the medical geneticist is prohibited by law. Should medical geneticists enter into a professional relationship with a potential counsellee who states from the beginning that he or she will not, under any circumstances, contact relatives and that a genetic condition must be kept secret? It is ill-advised to permit counsellees to dictate the terms of communication, especially in situations where harm to others may well be a factor. Absolute confidentiality cannot rationally be promised in all medical relationships. A better approach is not to promise absolute confidentiality at the outset of any genetic counselling, since the duty to inform others at risk will take precedence over any presumed right of the counsellee to keep the risk a secret. Physicians can make it clear to a counsellee that if the counsellee will not carry out his or her own duty, this refusal places the physician in an intolerable position. If a history of alienation and emotional problems in the family emerges, the counsellee can be offered help from a mental health specialist with the task of disclosure.

7.2.3 Monozygotic Twins: A Special Case

Monozygotic twins are individual, unique human beings who share the same genes. Each should first of all be respected as an individual person with individual needs, opinions, hopes, and desires. However, because genetic testing of one individual will inevitably reveal the status of the other, both should come to an agreement about whether to be tested before the medical geneticist proceeds with testing. If, after extensive counselling, they cannot agree - one wishes to be tested and the other does not wish to know the results of the test - the physician should proceed to test the person who requested it, provided that (1) both parties are fully aware of the possible consequences of testing one individual; and, (2) the party who does not wish to know the test result of her/his twin has had sufficient time and opportunity to protect him/herself against learning the result inadvertently. To refuse to test the twin who requests it would be to deny the uniqueness of that twin as a human being.

7.2.4 Spouses/Partners

Confidentiality should be overridden in only the most serious cases (and only if not prohibited by law) because of potential damage to the marriage and to its living children. In some cultures the woman is blamed to greater extent than the man for reproductive failures of all kinds. Therefore in cases where the woman is found to carry an autosomal dominant disorder, a balanced translocation, or an X-linked disorder, the counsellor should weigh carefully the benefits and harms of disclosure to her spouse.

Disclosure situations are of three types:

- If a couple intends to have children, individuals should share information with their partners in order that both be aware of potential harms to a future child. Professionals have a moral obligation to remind their counsellees of this. Doctors should be permitted to tell spouses/partners without the counsellee's consent, if children are contemplated, according to the guidelines for overriding confidentiality when other family members are at risk, a future child being considered a family member (see 7.2.2 above).

- A counsellee's genetic condition affects the spouse's future. Even when children are not intended, a family history or a diagnostic or presymptomatic test may have important bearing on the marriage. An example is a family history of Huntington's chorea, which may require the spouse to provide ten or more years of constant care for the affected partner. The spouse or partner deserves an explanation, even if there is no risk of genetic harm to the spouse. Ideally, the time to inform is before marriage, if the information is available. After marriage, the medical geneticist's concern is whether disclosure might destroy a marital relationship begun under a different set of assumptions. The approach in this case ought to follow the approach that most medical geneticists take to the incidental finding of nonpaternity. The information is primarily the counsellee's and the counsellee should be offered help with the emotional and ethical dimensions of the decision about disclosure to a spouse. The risks of not telling a spouse involve harm to a marital relationship grounded in promises of mutual support and trust. A secret of this magnitude is not likely to be kept without damage to the relationship itself. However, since there is no risk of direct genetic or physical harm to the spouse from non-disclosure, there is no ethical reason for geneticists to consider a breach of confidentiality. Medical geneticists should encourage the counsellee to consider the benefits of full disclosure and to seek help if there are emotional problems. However, if disclosure causes a threat to the marriage, the physician may support a decision not to disclose.
- Nonpaternity. Ideally, the counsellor should prevent the situation of disclosure of an incidental finding of nonpaternity by telling the woman alone, before testing, that the test could reveal nonpaternity. The woman may then decide to withdraw from testing. In practice, it may be difficult to counsel a woman alone in some cultures. In that case, she can neither be warned in advance nor be told of an incidental finding of nonpaternity.

Medical geneticists should keep in mind the well-being of the entire family and should remember that in many societies the woman is vulnerable to physical, social, psychological, and economic abuse. Often the geneticist does not know the history of a family's sexual interactions or whether these were voluntary or coerced. Therefore it is inappropriate to pass moral judgements on nonpaternity. There is probably never a justification for a physician to reveal incidental findings of nonpaternity to a husband. Usually it is sufficient, for purposes of providing information relevant to future childbearing, to tell the mother alone, without the husband/partner present. How she uses this information will depend upon the culture and herself. If the social or psychological environment may permit the mother to tell her husband/partner without undue harm to herself or the child, the counsellor should describe potential psychological benefits of disclosure, including relief from the burden of keeping a secret and greater honesty in family relationships. Decisions about whether to tell the husband/partner should be the mother's alone, however, after full discussion of physical, psychological, social, and economic consequences. If the mother decides to tell her husband/partner, the counsellor should stand ready to provide psychological and social support, including referrals to sheltering agencies.

Information about nonpaternity should be disclosed to the mother even if the couple is no longer capable of having children and there is no genetic risk. The information may have important bearing on family interactions and therefore should be known to at least one member of the family. If it is not possible to see the mother alone, it is better not to provide the information to anyone than to risk harm to her and to the child.

If a husband or partner asks directly whether he is the father of a child, the geneticist should follow the principle of preventing harm to the mother.

7.2.5 Non-Biological Relationships other than Nonpaternity

Sometimes incidental findings reveal non-biological relationships (other than false paternity) within a family, e.g., false grand-paternity or undisclosed adoption. The geneticist should proceed on the general assumption that at least one person knows about this non-biological relationship and if it is possible to identify and locate that person, should discuss the finding with that person and offer counselling about the benefits and harms of wider disclosure. The purpose of disclosure to at least one person is to help in understanding of screening or testing results and to enable that individual to decide about disclosure to other family members. If no living person exists who would be aware of a non-biological relationship, the geneticist should not disclose this relationship unless disclosure is required to prevent serious genetic harm to living or future persons.

7.2.6 Employers and Insurers

Third party access to genetic information is an issue that already receives great public attention. The new genetics may reveal asymptomatic conditions that may manifest themselves only at mid-life or in old age. The new genetics also reveals susceptibilities or risks for developing common diseases such as heart disease, breast cancer, or diabetes. These are risks, but not certainties. Information about future risks in healthy persons may be entitled to special privacy.

Any discussion of insurance should separate health insurance, which is a form of health care financing found in some countries, and which usually includes a profit motive, from life insurance or pensions that are based on insurance principles. Health care should be a basic human right, independent of ability to pay and devoid of profit motive. Only in the context of health care for all will access to genetics services be just and fair. Health care should be provided to all, regardless of genotype. In a just and ideal health care system, there may be no need to conceal genetic information from those who finance health care. However, as long as private health insurance and pensions based on insurance principles exist, there is a need to strictly protect the privacy of individuals (Berg, 1984; Berg and Fletcher, 1986).

Life insurance, unlike health care, is not usually considered a basic human right (though government-financed social and economic support for families of the deceased may be) and discussions of life insurance should be separate from discussions of health care. Life insurers and employers argue that much genetic information is already available from family medical histories and that they have been gathering this type of information routinely for years. This does not mean, however, that existing practices are entirely ethical (National Institutes of Health, 1993).

Most medical geneticists around the world agree that employers, life insurers, and other institutions, such as schools, should not have access to an individual's test results without the individual's consent (Riskin and Reilly, 1977). Consent, however, offers no free choice if an employer or school has the power to coerce consent by withholding employment or school admission. Insurance is based on the principle of sharing unknown risks. Therefore genetic testing or genetic information should not be a precondition of any kind of insurance, including a reasonable amount of life insurance. In at least one nation -- the Netherlands -- laws specify a near-universal right to a minimum amount of life insurance, regardless of risk.

There are two basic approaches to protecting individuals in the area of employment. The first is to protect privacy by making access to information about an individual impossible, even with that individual's consent. Some countries are already following this approach for insurance. According to this view, employers should be prohibited by law from requiring presymptomatic tests or susceptibility tests as a condition for employment, and prohibited from refusing employment to persons at known genetic risk or favouring persons with a "desirable genetic test result". If an individual decides to be tested, employers should be prohibited by law from access to test results, even if they paid for the test and even if the worker gives consent. If a fetus has been tested and carried to term, prospective employers should have no access to the child's test results.

The second approach is to allow access to information but prevent its being used for a discriminatory purpose. Many have pointed to the need for laws to protect, not privacy, but basic human rights to health care and employment (Billings et al, 1992; Natowicz et al, 1992). This is best done by extending legislation protecting those with disabilities to include persons with mutant genes or genetic predispositions to multifactorial disorders.

Jobs involving public safety (see 6.5 above) may perhaps be a (relatively rare) exception.

7.2.7 Other Institutions

Schools

Schools may have a valid interest in learning about a child's genetic status if a precise diagnosis will be useful in planning the child's education. Medical geneticists should guard such information conscientiously and reveal it to a school only if it will demonstrably be used to help in planning an improved educational programme for the child and only with the consent of the parents. Results of any tests that are presymptomatic for later-onset disorders or for carrier status should not be revealed to schools, in the interests of preventing discrimination.

Adoption agencies

Adoption agencies should not be permitted to ask prospective adopting parents about their genetic status, except insofar as this is directly related to their ability to care for a child while the child is still a minor. Risks for parental disorders that may occur far in the future or that are not relevant to the child's care should not be revealed to adoption agencies.

Motor vehicle agencies

Agencies that license drivers should have access only to information directly relevant to ability to operate a motor vehicle.

7.2.8 Government Agencies

Centralized record-keeping offers benefits to patients and medical researchers and also allows recontacting of individuals and families in the event of new medical discoveries. Any such registries should be in the hands of clinicians, not governments, and should be protected by the strictest standards of confidentiality (National Health and Medical Research Council, 1992; Harper, 1992). Such registries have made possible the location and treatment of women with PKU to prevent maternal PKU. Data collected can be used to monitor changes

in incidence, effectiveness of screening programmes, and quality of genetic laboratory services.

7.3 Methods for Protecting Privacy

Medical geneticists must constantly be aware of threats to individual privacy (Privacy Commissioner of Canada, 1992). Many of these come from ordinary sources, such as multi-line telephones or fax machines. Professionals should be aware that medical records, including family histories, typically pass through many hands. Information not directly relevant to a person's genetic status should not be entered into a family history. For example, it is not appropriate, in constructing a family history for thalassaemia, to note that the counsellee's uncle spent time in jail. The family history ideally should contain only information directly relevant to the counsellee's disorder, symptoms, or carrier status. Since the genetic component of most behavioural conditions is not yet scientifically established, geneticists should take particular care in protecting the confidentiality of behavioural reports. Because others may be able to identify individuals in a large kindred (even without names), especially in small communities, it is important to restrict the family history to those individuals whose information is relevant to the index patient. Counsellees should have the opportunity to see or hear all information about them that will be referred to other physicians. They should also be able to understand and approve the non-technical aspects of this information.

Information about identifiable individuals should not be transmitted by telephones with multiple extensions, by fax machines with more than one user or kept in an office open to more than one person, by electronic mail with a common password, by postcards, or by persons who have not been instructed about the importance of confidentiality. Records of identifiable individuals should not be kept on open shelves or in computers with a common password.

The confidentiality applied to records should also apply to the fact of a person's having visited a genetics clinic. Some people do not wish it known that they have visited a clinic. Clinic appointments or follow ups should not be sent out by postcard. Return addresses on envelopes should not refer to a genetics or prenatal clinic. If appointments are made by telephone, members of the clinic staff should apply specific procedures to be sure that they speak only to the party concerned. Medical information should not be given to a person calling the clinic by telephone unless the person is known to the counsellor and has previously received genetic counselling. Names of counsellees or relatives should not be provided to third parties without the counsellee's explicit request or consent, and should not be provided to commercial entities at all. Similar safeguards should apply to all recipients of medical care, not only those at genetics clinics.

8. Presymptomatic and Susceptibility Testing

8.1 Definitions

Presymptomatic testing (e.g., for Huntington disease) identifies individuals who will develop a genetic disorder if they live long enough. Susceptibility testing identifies persons who are at increased risk for developing common diseases, such as heart disease, but who may never develop the disease in question.

8.2 Benefits and Risks

8.2.1 Benefits of Testing

Medical benefits

In some cases, presymptomatic testing (e.g., for familial polyposis coli) can lead to prevention of the disorder's most serious effects (e.g., by colon surgery to prevent cancer.) Susceptibility testing can lead to preventive programmes for heart disease or intensive regular examinations that make possible early diagnosis and treatment (e.g., for breast cancer).

Life-planning

In other cases, where successful prevention or treatment are not possible, as in Huntington disease, the major benefit of presymptomatic testing is to provide information for planning one's life and for deciding whether or not to have children. For many people, life-planning is a major reason for seeking testing. Whether test results affect life plans (including reproductive plans) will depend upon:

- the risk given;
- the age of onset of the Mendelian disorder or common disease;
- the length of time between the test and the probable age of onset;
- perceived severity of the disorder or disease;
- the availability of support systems for people with the disorder or disease; and
- personal and cultural values and perceptions of disability.

Social Planning

Marshalling social support is another putative benefit of testing. At least in theory, societies could use the results of presymptomatic tests to plan adequate financial and physical support for persons who may develop disabilities and could use anonymous epidemiological data from susceptibility testing for public health planning.

8.2.2 Risks of Testing

Risks include depression, breaches of confidentiality, disruption of family life, loss of job and health care, and social stigmatization for those whose tests are unfavourable or indicate an increased risk. Risks also include depression and "survivor's guilt" for those whose tests are normal.

8.3 Recommendations for Offering Tests for Susceptibility to Common Diseases

Testing of Individuals with a Family History

Genetic testing of persons with a family history of heart disease, cancer or other common preventable or treatable diseases that may be of genetic origin should be encouraged, in order to identify persons at elevated risk and to institute preventive or surveillance measures (Berg, 1994). Testing should be voluntary.

Population Screening

Population screening should only be done for purposes of disease prevention or early diagnosis and treatment. It is unethical to screen for disorders that cannot be treated or prevented. Participation in screening should be voluntary.

Recommendations for Offering Testing or Screening for Susceptibility to Common Diseases

Susceptibility testing or screening should be available for adults who want it, provided that confidentiality can be guaranteed. Employers, health insurers, schools, or other institutions should not know that a person has been tested, should not have access to results of tests, even with the person's consent, and should be legally enjoined from attempting to coerce individuals to reveal test results.

8.4 Recommendations for Offering Presymptomatic Tests

Presymptomatic testing should be available for adults who want it, provided that the following conditions are met:

- Confidentiality can be guaranteed. Employers, health insurers, schools, or other institutions should not know that a person has been tested, should not have access to results of tests, even with the person's consent, and should be legally enjoined from attempting to coerce individuals to reveal test results.
- The person to be tested is fully informed about the limitations of testing, including the possibility that tests may be uninformative, that they may provide mid-range probabilities that are not close to 0% or 100%, that in any case they do not provide absolute certainty, that they do not predict exact age of onset, and that (for some disorders) they may not predict severity of symptoms.
- The person is not mentally ill at time of testing.
- There is evidence that the information provided by testing would be used to prevent harm to counsellee, spouse, family, prospective children, or others.
- Testing is accompanied by a counselling programme appropriate for the disorder. For some disorders, one session may be sufficient. At the other extreme, a severe disorder such as Huntington's chorea may require, as an ideal, three or four pre-test counselling sessions, unlimited follow-up sessions for those with unfavourable test results, and a follow-up session for those with normal results to alleviate "survivors' guilt".

A flexible approach, gauged to the needs of individuals, is preferable to a protocol specifying a certain number of sessions. If married, counsellees should be counselled together with their spouses for some of the sessions. This is especially important if children are contemplated.

9. Testing Children and Adolescents

9.1 Guidelines for Testing

The following guidelines for genetic testing of children take into account the increasing respect for minors' autonomy in the overall context of medical care (Wertz et al, 1994). There are four general types of situations in which testing may be requested.

- Testing for conditions for which treatment or preventive measures are available. Examples are familial polyposis coli, where removal of the colon in the teenage years may be necessary to prevent cancer, and severe familial hypercholesterolemia, where diet and medical treatment reduce cholesterol levels. For such disorders testing of minors is tantamount to diagnosis and should proceed according to consent guidelines established for other necessary medical interventions (Holder, 1977, 1988, 1989; Nicholson, 1986). Testing should be offered at the earliest age when health benefits accrue, but need not be offered before this time.
- The test has no health benefits for the minor, but may be useful to the minor in making reproductive decisions in the near future. Examples are carrier testing for autosomal or X-linked recessive disorders (e.g., cystic fibrosis or fragile-X syndrome), or presymptomatic testing for adult-onset disorders (e.g., Huntington's chorea). If the law permits testing of minors, the minor should be the primary decision-maker. Professionals should probe to discern whether the minor is acting on her/his own behalf (perhaps in agreement with parental suggestion), or is merely carrying out parental wishes without actually desiring to be tested. Minors should have the "negative right" of not knowing about their genetic status at all if they so desire (Clarke, 1993; Beauchamp and Childress, 1994). Ordinarily, testing will not be warranted unless either the minor or the minor's partner has a family history of a disorder.
- There are no medical benefits and no current reproductive benefits from testing, but parents or minor request it. Examples include parental requests for cystic fibrosis carrier testing of their children or Huntington's chorea testing on children who are well below reproductive age or who are not contemplating reproductive activity in the immediate future.

Testing in the absence of medical benefit or current reproductive benefit is best avoided. It is not "necessary" medical care and does not relate to reproductive rights.

The age at which the emotional and legal maturity required for consent appears is highly variable and also depends on the seriousness of the genetic disorder. Most often it will be advisable to defer testing until adulthood. If no clear benefits exist, parents should restrain their desire to know, and physicians should not yield to their request.

There is ordinarily no ethical justification for testing minors in the absence of proven medical benefits.

Decisions that override parental autonomy may be necessary in order to prevent harm and to preserve a minor's future autonomy, which should be the paramount considerations. Actions that place parental autonomy above all other concerns may lead to harm (Engelhardt, 1982; Thomasma 1983; Brett & McCullough, 1986). For example, a parental request to test a three-year-old for adult polycystic kidney disease

or a seven-year-old for predisposition to familial Alzheimer disease provides no medical benefit to the child and may lead to stigmatization.

- Testing is carried out solely for the benefit of another family member. This occurs frequently in DNA linkage analysis, where several members of a family, both affected and unaffected, must be tested in order to find out whether a particular individual (or a fetus) has a gene. Sometimes small children must be tested in order to enable their parents to use prenatal diagnosis in the next pregnancy. Such testing has a clear medical benefit, but not to the individual tested. In all cases, the test should have a clear usefulness for others, and the rationale for the test, including the name and description of the disorder (but not the name of the person on whose behalf the minor is to be tested, except with that person's permission) should be explained, insofar as possible. The minor should have the opportunity to decide, upon reaching adulthood, whether to know or not know the results.

9.2 Children Awaiting Adoption

The approaches suggested for parents' biological children should also apply to adopted children and children awaiting placement for adoption (Morris et al, 1988). Testing a child for untreatable adult-onset disorders prior to adoption makes the child into a commodity undergoing quality control.

9.3 Conflicts between Parents

Parental conflicts over testing pose another problem. Care should be taken, however, to avoid placing an undue psychological burden on the child. If testing provides a medical benefit or testing is done on behalf of other family members, it seems appropriate to side with the parent who wishes testing if treatment is necessary immediately, and to work toward resolution of the conflict if treatment can be postponed. An objective hearing by a standing review committee, established by the clinic for this purpose, would help to mediate disputes within families.

9.4 Disclosure of Test Results to Children

It should not be assumed that parents will convey full and accurate information years after a test is performed (Fanos and Johnson, 1992). Parents have an ethical obligation to convey the results of the tests to children at such time as the child can understand and benefit from the information. Professionals have an obligation to establish information networks that may enable them to follow families as they move, so that the professional can recontact children when they reach adulthood in order to make sure that they receive their test results. In order to make recontact possible, the test results should be placed in the child's primary care record for the information of subsequent physicians.

10. Behavioural Genetics and Mental Illnesses: Dangers of Stigmatization

Genetic and biochemical factors probably contribute to many behavioural disorders and mental illnesses, including alcoholism and schizophrenia. With the exception of single-gene disorders such as Huntington's chorea, however, the genetic contribution is usually only one among several causative factors. Biochemical factors predisposing toward illness are not necessarily genetic; they may originate during pregnancy, as a result of maternal exposure, or they may originate after birth as a result of bacterial, viral, or chemical exposure.

Biochemical predispositions may also result from the effects of malnutrition, both before and after birth, or from a childhood marked by prolonged or repeated anxiety (e.g., living in a war zone).

Genetics professionals have a social obligation to prevent or minimize stigmatization attaching to behavioural conditions that the public regards as genetic or partly genetic in origin (Wertz, 1990; Andrews et al., 1994). Scientists should avoid presenting findings to the media that could lead to premature genetic explanations for common behaviours (e.g., violent crime, alcoholism). Genetic explanations should not lead us away from the essential task of creating a socially just and healthy environment for human development. Medical geneticists should take a strong public stand against using genetics as the sole or major explanation of social problems, such as violence or drug addiction. Cultures vary in their definitions of normalcy and deviance. Some behavioural conditions (e.g., severe schizophrenia) are widely recognized as illnesses; others (e.g., borderline personality disorders) may be socially accepted in some cultures. In order to minimize harm to those whose behaviour differs from the majority, it is generally best to use the broadest possible definition of normal. Usually a functional definition, such as "ability to love and to work" (Sigmund Freud), is the most practical. Such a definition assumes that the person can function independently and is not harming self, others, or society.

Prenatal tests for behavioural conditions that are accepted by some cultures but not others (e.g., homosexuality) would be inappropriate, if such tests were to become available. Application of such tests could lead to even more restrictive definitions of normalcy.

11. Adoption

Adoption should be treated equally with other means of family formation. Adopted children should receive the same treatment as biological children in the context of genetics services, insofar as possible. This means that adopted children, like biological children, should have access to the genetic histories of their biological parents, grandparents, and siblings, if relevant. Those responsible for the adoption should obtain and record the medical histories of both biological parents and may, if appropriate, transmit this information to the adoptive parents, taking care that only medical/genetic information directly relevant to the child's genetic health status is transmitted. The child's social background (e.g., conception as a result of rape, parent jailed for anti-social behaviour, parents promiscuous) is not part of a genetic history and should not be included in the medical information transmitted to adoptive parents. Such information is irrelevant if the child is a newborn or infant and only serves to stigmatize the child. A newborn or infant deserves a fresh start. (Information on social environment is relevant in adoptions of older children, but should not be part of genetic information.) Disclosure of a child's genetic background will help prepare the adoptive parents. In cases where a child is at high risk for a serious disorder that usually manifests in childhood or adolescence, or where family history may indicate risk of a behavioural disorder (one or both parents schizophrenic), it is best to inform the adoptive parents before adoption, so that they can decide whether they are able to cope with this risk. If they cannot cope, it is better that they forego the adoption of this particular child than that the adoption fail when the child is older. There is no need for adoptive parents to know about adult-onset disorders.

In general, the rules for testing children placed for adoption should follow those for testing biological children (see Part 9.2). Children should not be tested for later-onset disorders before adoption. Such testing makes an adoptive child into a commodity to be

prejudged on its quality. An exception may be made, however, in extraordinary cases if all efforts to place the child in the absence of testing have failed. No child should be without a family because of genetic risks. Such situations should be extremely rare; after careful counselling, it should be possible to find families who are willing to accept genetic risks. Testing any children before adoption sets a dangerous precedent that could make it more difficult to place children at risk in the future. In other words, testing one child could lead to testing many more, if families become aware that those responsible for adoptions will accede to their requests.

The health of an individual's biological parents, as they age, is a major predictor of that individual's own health and life expectancy. Adopted children, like biological children, should know major facts about the health of their biological parents as they age, and also the causes of their parents' deaths. There should be registers that provide an ongoing avenue for transmitting genetic information between the biological parents and the adoptive family (and the adopted child, when the child becomes an adult) if a diagnosis will be useful for prevention, diagnosis, treatment, or reproductive planning. Information about the child (e.g., diagnosis of a genetic disorder in childhood) that may be useful to the biological parents (e.g., in reproductive planning) should be transmitted to the biological parents, if possible, just as information about the biological parents is transmitted to the adoptive family. All such information should be anonymous, without revealing the names of biological or adoptive parents to each other.

Ideally, nations should establish confidential registers, regularly updated for changes of address, for transmitting medical/genetic information in cases of adoption or of procreation assisted by gamete donation. Adopted children should be notified of the causes of their biological parents' deaths. Persons who were adopted or who placed a child for adoption in earlier years should be informed of the existence of such registers and should be given an opportunity to enter information. The registers should include information about the health of siblings and half-siblings.

Such registers may also be used for transmitting names of biological parents and children, but only if both parties enter into the record a willingness to be contacted by the other. Willingness to be contacted should be verified, in writing, before a person's name is transmitted to the other party. Many adopted children do not search for their biological parents, even when records are open. Nevertheless, a child's desire to know the biological parents' identities should not supersede the parents' right to confidentiality.

12. Prenatal Diagnosis: Indications and Societal Effects

Prenatal diagnosis includes all methods of ascertaining the health of the developing fetus; biochemical screening (maternal serum alpha-fetoprotein, triple-marker screening), ultrasound, amniocentesis, and chorionic villus biopsy. New and experimental methods such as fluorescent in situ hybridization (FISH) technologies and isolation of fetal cells from maternal blood in the first trimester of pregnancy present no new ethical problems (WHO, 1992), and will not be considered separately. Prenatal diagnosis gives the couple information so that they can make plans for the future. It may also help the physician to prepare for a difficult birth. Ethical guidelines for the provision of prenatal diagnosis appear in Table 6.

Table 6

ETHICAL GUIDELINES FOR PRENATAL DIAGNOSIS

1. Equitable distribution of genetics services, including prenatal diagnosis, is owed first to those with the greatest medical need, regardless of ability to pay.
2. Prenatal diagnosis should be voluntary in nature. The prospective parents should decide whether a genetic disorder warrants prenatal diagnosis or genetic abortion.
3. If prenatal diagnosis is medically indicated, it should be offered regardless of a couple's stated views on abortion. Prenatal diagnosis can be used to prepare for the birth of a child with a disorder, provided that the risk to the fetus is minimal and is acceptable to the parents.
4. Prenatal diagnosis is done 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 sex selection, apart from sex-linked disorders, is not acceptable.
5. Prenatal diagnosis solely for relief of maternal anxiety, in the absence of medical indications, should have lower priority in allocation of resources than prenatal diagnosis with medical indications.
6. Counselling should precede prenatal diagnosis.
7. Physicians should disclose all clinically relevant findings to the woman or couple.
8. The woman's choices, including abortion or carrying an affected fetus to term, must be respected and protected.
9. Professionals who offer prenatal diagnosis have an ethical obligation to provide referrals for safe, affordable abortions, preferably within the nation, if the woman desires it after unfavourable findings.
10. Supportive counselling should be provided before and after genetic abortions.

12.1 Prenatal Diagnosis without Abortion

Prenatal diagnosis can be used to prepare for the birth of a child with a disability instead of making a decision about abortion. Some couples use it for exactly this purpose. As treatments for genetic disorders improve, there is less likelihood of abortion and greater likelihood that prenatal diagnosis will be used to prepare for the births of children needing treatment. The majority of medical geneticists regard this as a medically indicated use of prenatal diagnosis. Prenatal diagnosis should be offered to all pregnant women at elevated risk, regardless of their views on abortion. It is unfair not to offer prenatal diagnosis on the basis of an individual's or couple's views. Offering does not mean urging or coercing. It means simply presenting information about prenatal diagnosis.

Most women who request prenatal diagnosis in order to "prepare themselves for the birth of a child with a disorder" hope for negative results, so that they can continue the pregnancy with reduced anxiety. Reduction of anxiety among women at high risk is a

justified use of prenatal diagnosis. Helping couples to prepare themselves for the birth of an affected child provided that they understand and accept the risks to the fetus, is also an ethically accepted use of the procedure (Clark and DeVore, 1989), if the risk to the fetus is minimal. Some couples use the information from an unfavourable diagnosis to make early plans for treatment, housing, and education. Some couples change their minds about abortion after receiving unfavourable results. To refuse prenatal diagnosis is to prejudge a couple's behaviour. In fact it may be difficult to predict responses to an unfavourable result.

12.2 Equal and Affordable Access

Whatever prenatal diagnostic services exist in a nation should be available to all equally, regardless of ability to pay, as long as there is a medical indication for the service (see 12.4 below).

When genetic resources are scarce, medical geneticists should be able to prioritize their allocation in terms of (1) seriousness of the genetic condition, and (2) level of risk. In setting such priorities, medical geneticists should assume that most couples requesting prenatal diagnosis may be open to changes of mind after unfavourable test results, regardless of the couple's stated intentions.

Decisions made on the basis of prenatal tests should be the woman's. Counsellors should not make willingness to abort affected fetuses a precondition for receiving prenatal diagnosis. In fairness to parents who make different choices, health care systems should be prohibited by law from refusing maternity coverage if an affected fetus is carried to term, and should be required to cover the costs of medically indicated treatment for the affected child after birth. Different societies will necessarily have their own standards for reasonable care of affected children.

12.3 Effects of Differential Use by Different Social Groups

Not to offer services to all equally is not only unjust, but could lead to further social inequality as people of different social groups avail themselves of the services unequally. In many countries, women who have prenatal diagnosis tend to be better-educated and to have higher incomes than those who do not have prenatal diagnosis. The better-off and better-educated are using prenatal diagnosis at disproportionate rates to other classes. For example, in the United Kingdom, "The two-income family that has postponed child-raising until their mid-thirties would become the primary customers for chromosome analysis. This prospect challenges the British sense of fairness and the belief that health care is a right rather than a privilege." (Harris and Wertz, 1989).

The women who receive prenatal diagnosis today are not always the women at highest risk. The age distribution in childbearing suggests that poor women, without access to contraception, account for a disproportionate share of the births to women over 40. People from lower socio-economic groups are also at greater risk for exposures to environmental hazards, both at home and at work, that may cause fetal disorders. Although substance abuse and battering of pregnant women occurs in all social classes, these problems are less likely to receive consistent treatment among poor women.

In the future differential uses of prenatal diagnosis and genetic abortion by different social groups could lead to an unbalanced distribution of genetic disorders among social classes. "It will be the educated, articulate, vocal, and economically privileged who will use the system most effectively and for whom there will be the most marked fall in births of

affected children. Further, the burden of caring for handicapped children might increasingly fall on those who can least afford it and are least able to press for better services." (Harris and Wertz, 1989, p. 405).

12.4 Indications for Prenatal Diagnosis

The discussion below applies mainly to invasive and costly procedures (e.g., amniocentesis and chorionic villus biopsy) that are likely to provide a definitive diagnosis. By "indication" we mean a medical, psychological, or social rationale justifying the procedure.

12.4.1 Medical Indications

Pregnancies at Elevated Risk

Indications include all factors leading to elevated risk, such as advanced maternal age, family history of a genetic disorder, knowledge of an abnormal gene in the family, a previous child with a disorder, or suspect findings (i.e., ultrasounds in ongoing pregnancy). Many government commissions and professional bodies have agreed upon these standards. There is less agreement, however, about what disorders are sufficiently serious to warrant diagnosis.

Use of Prenatal Diagnosis for "Less Serious" Conditions

There is no universally agreed upon definition of "serious". Nor is there any definition of what may be considered serious in the future. Conditions and their consequences that were once frequently fatal in childhood (such as cystic fibrosis) are now medically treatable and more socially acceptable, and many affected individuals reach adulthood. Some individuals with Down syndrome hold jobs, albeit in protected work places. Most people who would once have been bedridden can now propel themselves in wheelchairs. People with hearing, visual, or motor disabilities can now enter many public buildings, apartments, and businesses, as the result of laws requiring accessibility. In other words, many disabilities are less "serious" than they were formerly, due to medical, legal, and social advances.

On the other hand, in many cases medicine has extended life without being able to treat the basic mental or neurological problems. Parents can grow old while still caring for an adult child with a mental disability.

Prenatal diagnosis reveals disorders that some medical professionals might not consider "serious", such as sex chromosome abnormalities, but which society continues to stigmatize. Some parents who want small families of one or two children may decide that a boy with XXY (Klinefelter syndrome) for example, is not the son they want. Although the boy will reach puberty with proper treatment, he will be infertile (a condition that many fathers associate, falsely, with impotence), may look different from his peers, and may have learning or behavioural problems. A couple may decide that they do not wish to invest their resources in this child if they could choose otherwise. Another example: a couple belonging to a social group that places a high value on a woman's ability to bear children may decide that a girl with 45,X (Turner syndrome) would be an economic burden. On account of her infertility, no one in that cultural group may marry her. Parents vary greatly in their perceptions of seriousness. What one couple finds acceptable, another may find extremely serious in terms of their personal expectations for the child, their culture's expectations, their economic situation, or their goals for their own lives (Ekwo et al, 1987). Although use of abortion may follow a range of perceived seriousness that starts with severe mental retardation (total

inability to communicate), early death, or extreme physical disability as the most serious (Wertz et al, 1991), a small percentage of couples might consider, for example, development of Alzheimer disease at age 60 a condition that warranted termination before birth, especially if they themselves had cared for a parent with Alzheimer disease. (Even though they might not be living to care for the child when the child reaches 60, they might consider the future suffering for the child extreme).

Following the principle of autonomy, physicians should respect the wishes of fully informed and counselled parents and let them decide what they consider serious, even if the majority of people would not agree with that decision. There are cultural as well as individual differences in how people define health and disease (Payer, 1988). Unless society is willing to raise the child, the decision is best left to the parents who will actually raise the child (Powledge and Fletcher, 1979; Juengst, 1988; Danish Council of Ethics, 1991; Cowan, 1992). Only they can define "serious".

It would be dangerous to create medical, legal, or social definitions of "serious", because these could infringe on couples' lives in several ways. First of all, a disorder now considered "serious", such as Down syndrome, could become less "serious" in its effects because of improved education and training. If Down syndrome were to be redefined as no longer "serious", anti-abortion activists could promote legislation making legal abortion after prenatal diagnosis difficult.

At the other extreme, a cultural majority could define a condition as "serious" when it is in fact treatable. This majority could enforce its views on people who hold minority views by refusing social supports for children with this condition. In order to accommodate minority as well as majority views in pluralistic societies, it is best to leave all such decisions to the parents, even if some decisions appear to be made on "frivolous" grounds. The alternatives to a parent-centred policy are: (1) to forbid any abortions after prenatal diagnosis, or (2) to allow abortions only for disorders where there is evidence that death or total neurological devastation shortly after birth would be expected. In the second alternative, society (or the government) would formulate a list of abortable disorders. The first alternative would force some parents to accept burdens that they are unable to bear. The second alternative is based on the view that the fetus and the newborn are equal. Most people around the world do not share this view. This alternative would impose one view (equality of fetus and newborn) upon all. It could also encourage pediatric euthanasia, if abortions are forbidden.

Accommodating all views, however, could leave the door open to some "cosmetic" decisions, for example, with regard to height and weight. Extreme variants in both weight and height are in a sense "medical" conditions and doctors would be ethically obligated to disclose major variations from the norm.

The best approach to prenatal diagnosis for so-called "less serious" conditions is to provide the most complete, unbiased education possible. This is especially important if parents have no experience with the disorder in question. What parents do after an unfavourable test result depends to a great extent on what the doctor, counsellor, or genetic support group tells them. For example, fewer parents decide to abort for sex chromosome disorders if provided with thorough, unbiased counselling (Holmes-Siedle, 1987).

Some parents will consider cystic fibrosis a "less serious" condition, especially as the media continue to report new treatments and hopes of cure. What the population at large does with carrier screening and prenatal diagnosis for cystic fibrosis will depend almost

entirely on what the media and the medical profession tell them. Most people have never seen anyone with cystic fibrosis. Parents who are told that children with cystic fibrosis are likely to die in their late teens after a long period of serious illness are likely to respond very differently to offers of prenatal diagnosis than are parents who are told that their child may live to 40 and have a productive life.

When a Woman's Partner Cannot be Tested

If a woman's test results identify her as a carrier of an autosomal recessive disorder and her partner cannot be found for testing, she should be offered prenatal diagnosis after full counselling about her risks of having a child with the disorder versus the risk of the procedure to the fetus. Withholding prenatal diagnosis in these cases would be unfair to the woman and her future child. Such cases should have lower priority in a health care system, however, than known carrier-carrier couples.

Sex Selection in Cases of X-linked Disorder

Sex selection is morally justifiable in some cases to prevent serious X-linked disorders that a healthy mother can transmit to her sons but not to her daughters. These include hemophilia and some forms of muscular dystrophy. A male fetus whose mother carries a gene for an X-linked disorder has a 50% chance of having the disorder. Some X-linked disorders cannot be diagnosed before birth. Identification of fetal sex and genetic abortion of male fetuses who are at 50% risk may enable the parents to prevent the birth of a child with severe medical problems. This use of prenatal diagnosis falls within ethically accepted uses of prenatal testing to prevent serious genetic disorders.

12.4.2 Maternal Anxiety

Maternal anxiety, in the absence of a known factor for elevated risk, is at the borderline of medical indications. In some nations with a large laboratory capacity it is considered a medical indication. In nations with limited laboratory capacity, it may be considered a waste of scarce resources. In deciding whether to perform prenatal diagnosis solely on the basis of maternal anxiety and mother's or couple's request, justice should be the primary concern. Unless public health resources are virtually unlimited, it is unfair to provide this service, because it means depriving others of some more needed service. It also poses an unnecessary risk to the fetus. Morbid anxiety in either parent, clinically confirmed by a psychiatrist or psychologist, warrants the service on humanitarian grounds. Sometimes this occurs in women who have cared for people with severe disabilities. A woman experiencing the usual anxieties of pregnancy, however, should not receive prenatal diagnosis solely on this ground.

12.4.3 Non-Medical Indications

These include (1) sex selection, in the absence of an X-linked disorder; (2) prenatal paternity testing; and, (3) tissue typing for possible organ donation after birth.

Sex Selection for Sex Desired by Parents

Two ethical issues are involved. The first is whether couples should be able to choose the sex of their children, and if so, under what conditions. The second is whether abortion is justified as a means to this end.

Direct requests for prenatal diagnosis for sex selection are likely to remain few in Western nations, in view of (1) the absence of a strong cultural preference for children of a particular sex; and, (2) personal and cultural objections to use of abortion for this purpose. Although the majority of North Americans believe that abortion should be available to others in a wide variety of situations, including sex selection, few would use it themselves (Wertz et al, 1991). Information about fetal sex is usually communicated to parents if they wish to know, though some clinics do not provide the information unless specifically requested (Hulten and Needham, 1987; Wertz and Fletcher, 1989b). Some countries have laws against divulging the sex of a fetus during the period in pregnancy when abortion on request is legal.

The major use of prenatal diagnosis for sex selection occurs in some developing nations where there is a strong preference for sons. There, a majority of prenatal diagnostic procedures are performed for sex selection rather than detection of fetal abnormalities. Ultrasound, although not always accurate, is affordable even to villagers and poses no known risk to the mother. In many nations of Asia, sex selection contributes to an already unbalanced sex ratio occasioned by neglect of female children. An estimated 60,000,000 to 100,000,000 women are missing from the world's population (Sen, 1989, 1990; Coale, 1991), including 29,000,000 in China and 23,000,000 in India. Whereas in the USA, UK and France, there are 105 women to every 100 men, and in Africa and Latin America the proportions of women and men are roughly equal, in much of Asia, including Pakistan, Afghanistan, Turkey, Bangladesh, India, and China, there are fewer than 95 women for every 100 men (United Nations, 1991). Families desire sons for economic reasons. In these nations, where most people have no social security or retirement pensions, sons are responsible for caring for parents in their old age. Daughters usually leave the parental family to live with their husbands and to help care for their parents-in-law. Even if a daughter stays in the parental home, she seldom has the earning power to support her parents. In some nations, a daughter represents a considerable economic burden, because her family must pay a dowry to her husband's family in order to arrange a marriage. A son's religious duties at the parents' funerals, although often cited as a reason for son preference, are of lesser importance than economic factors. These religious duties can be performed by other male relatives.

Ethical arguments in favour of sex selection in general, including pre-conception selection, are that (1) sex choice would enhance the quality of life for a child of the "wanted" sex; (2) sex choice would provide a better quality of life for the family that has the sex balance it desires; (3) sex choice would provide a better quality of life for the mother, because she would undergo fewer births and her status in the family would be enhanced; (4) sex choice would help to limit the population (Warren, 1985). According to these arguments, families that have the sex "balance" that they desire would be happier. Children of the "unwanted" sex, usually female, would be spared the abuse, neglect, and early death in childhood that is their documented fate in some developing nations (Verma and Singh, 1989; George et al, 1992), and that may occur to a less obvious extent elsewhere. Women would not be abused by their husbands for not bearing children of the desired sex. Women would not suffer repeated pregnancies and births in order to produce at least one child of the desired sex, usually a son. Couples would not have more children than they could afford in order to have a child of the desired sex. Many couples in developing nations would prefer to have at most two children. These couples could limit their family size and still have a son to support them in their old age, instead of continuing to have children until they have a son. The threat of world overpopulation might recede.

Each of the arguments above can be effectively countered. Arguments that sex selection will lead to a better quality of life for families, children, or women are comprehensible only in the context of a sexist society that gives preferential treatment to one sex, usually the male. Instead of selecting sex, societies should work to improve quality of life by making society less sexist. Although sex selection could prevent some abuse of unwanted female children and their mothers in the short run, it does not correct the underlying abuses, namely the social devaluation of women in many parts of the world and the gender stereotyping of children of both sexes in the rest of the world.

There is no good evidence that sex selection will reduce population growth in developing nations. Education of women in developing nations and increased opportunities for their employment outside the home are more effective means of reducing population growth than sex selection. In developed nations, sex selection will likely have no effect on population size, because most couples will not have more children than they wish in order to have a child of a particular sex (Dixon and Levy, 1985).

Arguments against all types of sex selection are based on the premise that all sex selection, including selection for the "balanced family" desired in some Western nations, helps to perpetuate gender stereotyping and sexism (Warren, 1985; Overall, 1987). Sex selection violates the principle of equality between the sexes (United States, 1983). In a nonsexist society, there should be no reason to select one sex over the other. Bayles (1984) has examined concerns that might be put forward for sex preference, including replacing oneself biologically, carrying on the family name, rights of inheritance, or jobs requiring either men or women. He points out that none of these reasons is valid. A child's sex does not make that child biologically any more "my" child than a child of the other sex. In modern societies, women as well as men can carry on the family name, inherit estates, and carry out most jobs. Conversely, men can care for children, elderly parents, or relatives with disabilities, tasks that usually fall on women and that could in the future lead to a preference for daughters. Warren (1985) points out that even in a nonsexist society, however, there would remain a desire for the companionship of a child of one's own sex. This is not a strong argument in favour of sex selection. Any activities that a parent can enjoy with a child of one sex, such as sports, vacations, or hobbies, can be enjoyed with a child of the other sex.

Another argument against sex selection is that it could increase gender inequalities, even in developed nations where parents usually prefer sons and daughters equally. Although these preferences are slight, there is evidence that in North America couples would prefer that the first-born be a boy or that they would prefer to have two sons and a daughter if they are to have three children (Pebley and Westhoff, 1982). Although there is no firm evidence that first-borns receive more economic advantages than later-borns (Warren, 1985) some social scientists believe that a society in which first-borns tended to be sons would tend to give more power to males.

There are additional arguments against sex selection if it takes place after conception. Prenatal diagnosis for this purpose is a misuse of costly, and in some nations scarce, medical resources. Sex selection negates the medical uses of prenatal diagnosis to detect serious disorders in the fetus and undermines the major moral reason that justifies prenatal diagnosis and genetic abortion - the prevention of serious and untreatable genetic disease. Using prenatal diagnosis to select sex could lead to a "slippery slope" toward selection on cosmetic grounds, such as height, weight, or eye, hair or skin colour. Some parents may select for such purposes, perhaps especially for weight (Wertz et al, 1991). Gender is not a disease.

Laws prohibiting sex selection would not necessarily prevent the practice, and could lead to further interference with reproductive freedom. A better approach may be to work toward equality of the sexes and against gender stereotyping, including the stereotyping of fetuses (Rothman, 1986; Sjögren, 1988), and to establish a moral climate against sex selection. Sex selection for non-medical reasons is not a medical service; doctors do not have to accede to counsellor requests or offer referrals. In cases where a doctor suspects that sex selection is likely to occur, he or she may consider withholding information about fetal sex until after the legal time limit for abortion has passed (and in some countries this practice has been established). The information is not related to the health of the fetus. The doctor should tell the couple the reason for withholding the information.

Prenatal Paternity Testing

In cases where paternity is uncertain, the woman or her partner(s) may request prenatal diagnosis solely for paternity testing. It is not clear whether withholding prenatal paternity testing would reduce or increase the number of abortions in situations where paternity is dubious. Withholding prenatal testing could increase interpersonal dishonesty. Openness is often the most beneficial alternative, especially in view of the child's future relationships with others. Each situation must be evaluated individually in the light of social, cultural, and family norms. Medical geneticists must acknowledge procedural risks to the fetus and should inform the woman as well as the man of these risks.

Prenatal paternity testing can also be used for forensic purposes, if pregnancy occurs after rape. In cases where the pregnancy may have resulted from criminal assault, it is especially important to know the truth about paternity so that the woman can make a decision about abortion. Probably few would question the use of prenatal diagnosis if rape or incest has occurred.

Tissue-Typing for Organ or Marrow Donation

Sometimes a couple with a seriously ill child wish to know whether their fetus, once born, will be able to serve as a donor of bone marrow or other organ transplants for the living child. Information about the fetus would enable them to make plans for the living child's future. This information, however, would also enable them to "save time" by aborting a fetus with an incompatible tissue type and conceiving another fetus that might have tissue suitable for a transplant. Professionals sometimes suspect that the latter motive underlies requests for prenatal diagnosis. Parents are understandably concerned over the health of their living child and deserve sympathy in these situations. They fear that time will run out before they can find a suitable donor. Nevertheless, if they are considering the fetus primarily as an organ donor, they are using that fetus as a means to an end rather than as an end in itself. A fetus should not be regarded as a tissue preparation for someone else, even if the transplant procedure may be harmless to the donor. Restraint would be strongly advisable in matters relating to tissue typing, because of the temptation that it provides to think of a fetus largely in terms of benefit to someone else. In order to prevent possible harm to the fetus, it is advisable to wait until birth with tissue-typing.

12.5 Effects of Prenatal Diagnosis on Societal Attitudes Toward People with Disabilities

Some people fear that increased use of prenatal diagnosis will shift social resources away from people with disabilities (Rothman, 1986; King's Fund Forum, 1987; Harris & Wertz, 1989; Schroeder-Kurth and Huebner, 1989), whereas others argue that no evidence

of this has appeared during the many years that prenatal diagnosis has been available (Motulsky and Murray, 1983).

In approaching this topic, it is important to remember that many birth defects are not purely genetic in origin. Common causes of birth-associated disability are prematurity, low birth weight and environmental exposure (Yankauer, 1990; WHO, 1992).

Altogether, chromosomal disorders (e.g., Down syndrome), single-gene disorders (e.g., Tay-Sachs, fragile-X syndrome), and developmental malformation syndromes account for about 43% of individuals with I.Q.'s under 50 (United States, 1979b; Andrews et al, 1994). Accidents at birth, prematurity, environmental or substance exposures, and unknown factors (possibly including some multifactorial genetic factors) account for the remaining 57%. It is important not to let the availability of genetic tests lead to the illusion that most disabilities are avoidable through prenatal diagnosis. Some fetal malformations cannot yet be diagnosed prenatally. Even disorders that can be diagnosed prenatally, such as Tay-Sachs, will not be tested for in low-risk groups and will continue to appear. Other disorders, such as neurofibromatosis, have a high new mutation rate. This means that disabilities will always occur, regardless of prenatal diagnosis. Society needs to be prepared to offer support to persons with disabilities. Even if every pregnancy underwent chromosomal prenatal diagnosis and testing for neural tube defects (an unlikely event, given the negative risk-benefit ratio for younger women) and every woman chose abortion of affected fetuses (also an unlikely event), children would still be born with genetic conditions or congenital malformations (unsuspected inborn errors of metabolism, new mutations, etc.).

Social and economic programmes to prevent prematurity and low birthweight should go hand-in-hand with public education about genetics and use of prenatal diagnosis. Prevention of disabilities - through adequate maternal nutrition, prenatal care, prevention of substance abuse or physical abuse, and prenatal diagnosis - is not at cross-purposes to increased support for living people with disabilities. It is illogical to argue that supports for people with disabilities will be reduced if there are fewer such persons. Much of the concern expressed by people with disabilities stems from the potential symbolic impact of widespread use of prenatal diagnosis on people's perception of disabilities in general. Public education about disability is one way of addressing these concerns.

The world is unlikely to have fewer persons with disabilities in the future. As societies age, we can expect more, rather than fewer, persons with disabilities of all types, including mental disabilities. It is therefore important to increase, rather than to contemplate decreasing, supports for persons with disabilities. It is also important to prevent any mandatory use of either prenatal diagnosis or its results.

Coercion should be avoided. There should be protection for the views of minorities who believe in the protection of all life. This does not mean that society should bear the costs of all aggressive life-support when treatment is ultimately futile; withholding such support is ethically permissible and is allowed by many world religions, although the degree of ethical stringency differs among them on this issue. The main point is that the availability of genetic tests must not be allowed to create an illusion that most disabilities are preventable and therefore unacceptable to society.