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Review of Ethical Issues in Medical Genetics

Report of Consultants to WHO
Professors D.C. Wertz, J.C. Fletcher, K. Berg

World Health Organization
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Preface

The international Human Genome Project (HGP) will rapidly make genetic information available on a worldwide scale previously impossible to imagine. All adults have a right, if they so choose, to know their genetic makeup and implications for the health of their potential offspring, to be educated about their own genetics, and to have the services available to act upon their knowledge.

The HGP, while not raising generically new ethical issues in medicine, exacerbates old ones, especially in regard to equitable access to genetic services, privacy, disclosure of genetic information, and freedom of reproductive choices. The HGP holds great promise for advances in human health but has also increased the public's concerns about genetics. To allay these concerns, to protect people and families with genetic disabilities, and to promote international cooperation, it is timely to discuss ethical issues in medical genetics and to propose guidelines on complex ethical issues for the providers of genetic services.

Within the next decade, newborn and carrier screening, and screening for common disorders such as heart disease, cancer, and neurodegenerative diseases, may greatly increase the role of genetics within primary health care. The inclusion of clinical genetics services as an integral part of basic health care should, therefore, be supported. All governments and their agencies related to delivery of health care need to examine the adequacy of current genetics services and how these can be improved in ethically acceptable ways.

This draft document reviewing ethical issues in medical genetics and genetic services in an international perspective and serving as background information relating to a shorter consensus statement "Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services" (WHO, 1998) was further revised by the original Consultants and prepared for publication. The entire content of the present version does not have the level of consensus among professionals that was reached in the *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* (WHO, 1998). Some ethical problems of medical genetics, e.g., abortion after prenatal diagnosis, choices about alternatives in assisted reproduction, and the status of the human embryo in genetic research, are highly debatable and, at this time in history, are issues beyond the reach of moral consensus among nations. It is also recognizable that the laws of nations differ with respect to these particular issues and that law is subject to debate and evaluation.

The recommendations in this document and in the *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* (WHO, 1998) are intended as points of departure for genetics professionals and public health officials to develop policies and practices in their own nations.

Part I
General considerations

Introduction : Importance of Genetics

Hereditary conditions affect millions of families throughout the world. About 5% of all pregnancies result in the birth of a child with a significant genetic disorder, congenital malformation or disability. An estimated 43% of cases of severe mental retardation (IQ <50) are caused by single genes or chromosomal abnormalities (Institute of Medicine, 1994). In developed nations, totally or partially hereditary conditions account for about 36% to 53% of paediatric hospital admissions (Institute of Medicine, 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, may have a genetic component (Holtzman, 1989).

Even though many affected individuals live full and happy lives and may not 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 for non-institutional, outpatient, educational, medical and social services, as well as lost economic output from family members who care for persons with genetic disorders. Therefore, continued efforts to develop effective treatments and make them available worldwide are important to the health of communities as well as individuals and families.

1. 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 and its application to all forms of life, from bacteria and viruses to plants, animals, and humans; (2) allocation and delivery of health care resources; (3) ethical problems that arise in clinical encounters between health care professionals and patients, and; (4) ethical problems in preventive medicine and public health.

1.1 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 of 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.

1.2 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 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 available 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 to prevent them, where possible.

1.3 Resources for Ethical Guidance

Approaches to medical ethics are varied. In describing the various approaches, we begin with principles-based ethics.

1.3.1 Ethical Principles in Medicine

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

Table 1. Relevant Ethical Principles in Medicine

- **Respect for the autonomy of persons**: respecting self-determination of individuals and protecting those persons with diminished autonomy.
- **Beneficence**: giving highest priority to the welfare of persons and maximizing benefits to their health.
- **Non-maleficence**: avoiding and preventing harm to persons or, at least, minimizing harm.
- **Justice**: treating persons with fairness and equity and distributing benefits and burdens of health care as fairly as possible in society.

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

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

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

The goals of justice can be described somewhat differently: treating persons fairly, giving persons what they deserve, or giving persons that to which they are entitled. The term “distributive” (or social) justice means to allocate benefits (e.g., property) and burdens (e.g., taxation) fairly and with equity, in order to enhance social harmony and cooperation.

Distributing the benefits (e.g., of diagnosis and treatment) and the burdens (e.g., rationing of expensive care or of research) of health care ought to be governed by ethically justified rules such as: to each according to need, to each according to an equal share or opportunity, etc.

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

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

WHO Member States should be encouraged to draw up public health policies that include standards for genetic services along the lines recommended in the Report of a WHO Scientific Group on Control of Hereditary Diseases (1996). People have the right to equitable access to genetic services according to the standard of care that exists in each country, according to need and, irrespective of the ability to pay. Some parties, such as women, children and people with disabilities, are especially disadvantaged and vulnerable in some societies and deserve special consideration. Professionals should help protect such persons wherever they are at risk of harm.

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

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

1.3.2 Knowledge and Use of Prior Cases (Casuistry)

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 that refers 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 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.

1.3.3 Professional Values, Relationships, and an Ethics of Care

When medical geneticists interact with the life histories and needs of individuals 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 neglects the fact that people are related both socially and genetically (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).

1.3.4 Asian Approaches to Bioethics

Asian approaches to ethics have always been based on relationships rather than on individual rights. According to bioethicist Renzong Qiu, "Rights-talk is a recent event compared to human history. In non-Western countries there has never been rights-talk before they met Westerners. Even now, in many third-world countries rights-talk is not so stressed as in Western countries, or has a different focus." (Qiu, 1998).

Asian approaches reject self-interest, individualism, and contractualism. The heart of Confucian ethics is love and care for others (Chan, 1964). Care for others is what differentiates a human being from an animal. Care for others is called *ren*, or humanness. According to Confucian thought, "Where do you come from? From your parents. So, filial piety is the beginning of humanness. Filial piety is a human being's first passion and first responsibility towards others. From this start you should extend your responsibility and passion to your sisters and brothers

(fraternity), your children (kindness), your spouse (fidelity), your friends (sincerity), your patients (compassion), your countrymen (harmoniousness), and foreigners (peacefulness). Because the birth and development of an individual owes so much to others, he has the duty to care for them. Care for, concern with, and responsibility towards future generations and toward the disadvantaged or vulnerable is a natural extension of *ren*, humanness." (Qiu, 1998) This sense of responsibility is not based on rights of other individuals, but on "consciousness of humankind as a whole." Human relationships exist because we are all part of humankind, not because we have contracts between individuals.

In practice, this ethic means that "genetic services should be approached more humanistically (by a warm heart) rather than scientifically or technically (cool head)" (Ohkura 1996). Health care providers should be more than technicians or providers of information. The family must be understood in much broader terms than in the West, and the health care provider must think in terms of relationships among family members rather than in terms of conflicting "rights" of different parties.

Asian bioethics tends to support new genetic technologies. "According to Chinese tradition, man could become a Sage, A True Man, a Buddha, that is, man could become infinite through his or her creative practical activities." (Lee 1998). The Tao (Way of Heaven and Earth) is immanent in every human being in daily activities; humans form a trinity between Heaven and Earth. Overcoming defects is realizing Tao to the full. Therefore "Confucianism supports the development of biotechnology in the relieving of human defects, including use of alternative reproductive technologies" (Lee 1998).

Some Asian bioethicists also support enhancement. According to Sakamoto (1996), the Buddhist Law of Eternal Change means that "nature" is not a stable, fixed entity; therefore no line can be drawn between the "natural" and the "artificial". Human beings' attempts to better themselves are in accord with the Law of Eternal Change, rather than a violation of some eternally fixed "human nature". Other Asian commentators, however, claim that enhancement through germ-line gene therapy would be a denial of our responsibility towards future generations (Qiu 1998).

1.3.5 Reconciling Western and Asian Ethics

Although Western Ethics is based on rights and principles and Asian ethics is based on caring and relationships, often the practical outcomes of the two approaches are similar. Experienced Western genetic counsellors know that they cannot base their practice entirely on individual rights and autonomy. In fact, in the United States, the National Society of Genetic Counselors (1993) has a Code of Ethics based entirely on relationships rather than on principles. Conversely, Western principles of non-maleficence, beneficence, and justice are implicit in the Confucian ideal of humanness. The difference between Asian and Western ethics lies principally in the amount of credence given to the autonomy, privacy, and "rights" of atomized individuals.

This document uses Western principles-based language in the tables, primarily because it is easier to generalize a set of actions on the basis of principles than on the basis of relationships. The discussion sections point to the importance of relationships and of empathy between counsellors and families but, it would be difficult to make suggestions or recommendations on the basis of relationships alone. Relationships-based ethics tend to dissolve more general situations in medicine into individual cases.

1.4 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 the potentially adverse effects of screening, diagnostic testing, and experimentation. Professionals should serve as advocates for such persons wherever there is a possibility of harm.

1.5 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. An important reason for offering prenatal diagnosis to all is that sometimes treatment may be possible for the fetus, or the procedures used at delivery may be planned for the best outcome.

Respect for freedom of choice does not mean that all technically possible services must be provided at the request of individuals and families, but only that services normally provided be provided equally, without regard to people’s 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 and 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.

2. Genetic Services in Ethical and Social Context

2.1 Goals and Practices of Medical Genetics

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

Conditions studied by medical geneticists include diseases caused by defects in single genes (e.g., haemophilia, sickle cell anaemia, cystic fibrosis), disorders caused by interaction between several genes and environmental factors (e.g., common congenital malformations, diabetes, hypertension, cardiovascular disease, breast cancer, mental disorders) and conditions caused by chromosomal anomalies (e.g., Down syndrome). Diagnostic work in medical genetics includes laboratory work at the DNA, protein, and chromosome levels as well as clinical observation of disorders, including birth defects. Whereas single-gene disorders are rare, conditions caused by an interaction between genes and environmental factors are frequent and include disorders such as cardiovascular diseases, several cancers, asthma, diabetes mellitus, and mental disorders. Preventive aspects of work in medical genetics includes identification of high-risk individuals with respect to common disorders for the purpose of preventing disease (e.g., heart disease) or securing early diagnosis and treatment (several cancers). At present there are significant research efforts aimed at developing somatic cell gene therapies or therapies to block the function of genes.

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

2.2 Application of Ethical Principles to Genetic Services

The application of the ethical principles to genetic services described in Section 1 is illustrated in Table 2.

2.3 Facilitating Individual/Couple Choices Regarding Parenthood

2.3.1 Freedom of Choice

Promoting freedom of choice is essential to the goals of genetics. Close to 100% of 682 medical genetics services professionals[1] in a 1985 survey in 19 nations said that the following were

[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.

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. 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 and 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; it 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 others in order to lessen their anxiety.

Table 2. Ethical Principles Applied to Genetics Services

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

2.3.2 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 child rearing. Usually this means the family, which takes many forms around the world. The Asian concept of family is broader in its range than in most Western nations. Many people may be responsible for the support and care of a child during its lifetime. Decisions about whether to have a child or to carry a fetus with a genetic condition to term should involve both parents, but the final decision should be the woman’s. In cultures where extended families play an active role in care, support, and nurture, opinions of other close relatives may play a role but should not override wishes of the parents.

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. The option of giving up a child for adoption should also be considered if the parents are unable or unwilling to care for a child.

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 the people they counsel to work toward their own decisions. Professionals should be careful to avoid the force that the "technological imperative" (belief that availability of a procedure generates a moral imperative to use it) may exert on decision-making.

2.4 "Is Medical Genetics Eugenics?"

2.4.1 Meanings of the term "Eugenics"

The word "eugenics", from the Greek for "good birth", was coined in 1883 by Sir Francis Galton, an eminent British scientist. Although the term still carries its original Galtonian meaning of "healthy birth" in some parts of the world, it is usually employed as a pejorative today, without careful attention to its meaning. Recognizing that genetics aims to improve the lives of individuals and families (but not to "improve" the genetic health of the society), that in human populations there are no "superior" or inferior" genomes, and that human diversity contributes to the survival and richness of humanity, it is important that the genetics profession undertake an examination of 1) the meaning of eugenics, both historically and in the modern world; and 2) whether existing or future practices may constitute or lead to eugenics.

Most modern authors associate eugenics with Nazi programs to eradicate Jews, Gypsies, homosexuals, and other "inferior" groups, in other words, with genocide. In fact, eugenics was transpolitical, spanning the entire spectrum from ultra-conservative to ultra-radical. In the United Kingdom, most major social reformers and liberals considered themselves eugenicists. John Stuart Mill, the best-known proponent of utilitarianism, playwright George Bernard Shaw, and philosopher Bertrand Russell were all eugenicists, though none believed in coercion by the government.

We prefer the following working definition of eugenics: "A coercive policy intended to further a reproductive goal, against the rights, freedoms, and choices of the individual." For purposes of this definition, "coercion" includes laws, regulations, positive or negative incentives (including lack of accessibility to affordable medical services) put forward by states or other social institutions. Cultures or medical settings may be implicitly coercive and are aware of the need for vigilance against tacit coercion, but considered such problems as part of the general social context rather than as eugenic programs.

Under the above definition, knowledge-based, goal-oriented individual or family choices to have a healthy baby do not constitute eugenics. Such choices are unlikely to affect the gene pool or to reduce the numbers of persons with disabilities. Most disabilities are not the results of chromosomal or single-gene disorders, and most babies born with a genetic disorder are born to families with no known risk for having a child with that condition.

Eugenics is directed against whole populations, whereas the work of today's clinical geneticists is directed towards individuals and families. However, it is important to be aware that collective results of individual decisions could lead to social policies that discriminate against the minority who make different decisions and especially against persons with disabilities. In a democratic society this result could occur by virtue of majority vote to restrict services.

2.4.2 Public Health Practices

Several terms and practices relevant to public health may be wrongly confused with eugenics. "Euphenics" means the improvement of the phenotype by biological means. The term was proposed by the Russian biologist N.K. Koltsov, who published an article under this title in a 1929 Soviet medical encyclopaedia, and formulated independently in the 1960's by J. Lederberg. Essentially, euphenics involves the incorporation into preventive and therapeutic medical practice, of the broad advances that are being made in molecular biology, immunology, neurophysiology, and other rapidly growing biological fields. Lederberg, in particular, has been a strong advocate of euphenics as "a corrective measure for our genetic ills." (Lerner and Libby 1976, p. 385).

Euphenics is basically good health care. State-mandated newborn screening programs to identify and treat newborns for conditions where early diagnosis and treatment benefit the newborn are not eugenic programs, because their primary purpose is to help the newborn. Reproductive information and counselling for the parent is a side effect of state programs, but is conducted on a voluntary basis (American Society of Human Genetics, 1999).

"Euthenics" (Lerner and Libby, p. 385) is "improvement in the environment." A good example is government-required warnings on alcohol and cigarette containers that drinking or smoking while pregnant may harm the fetus. These warnings do not legally restrict a woman's activities, but attempt to improve the environment for the fetus. Adding iodine to salt (to prevent thyroid deficiency), vitamin D to milk (to prevent rickets), or folic acid to cereal products (to prevent spina bifida) are other examples, as is vaccinating women for rubella to prevent rubella in the fetus (rubella may damage the fetus).

2.4.3 Eugenics in History

Most nations have a history of eugenic thought or practice. Some have tried to keep gene pools separate by forbidding legitimate unions between members of different social groups. The caste system in India represents perhaps the largest "eugenic" experiment ever, spanning almost 3000 years (Dobzhansky, 1973). Anti-miscegenation laws prohibiting marriages across racial lines in U.S. southern states made a similar attempt (Lawson, 1995). Such programs caused much social discrimination but inevitably failed to alter gene pools.

Immigration laws also attempted to restrict gene pools. The U.S. immigration law of 1924 was aimed at preventing immigration of Asians, Africans, and Southern or Eastern Europeans, partly on the basis of behavioural genetic studies purporting to show that these groups were inferior. In the United States, sterilization laws attempted to stem a purported threat to the gene pool from poor whites living in rural areas, a group that could not be kept out by immigration laws or kept in place by segregation laws. About 30 states passed laws requiring sterilization of "imbeciles", "feeble-minded," epileptics, mentally ill, criminally insane, etc. Between 1907 and 1960, at least 60,000 people were involuntarily sterilized (Reilly 1991). Most of these people were in institutions and most advocates for sterilization were behavioural psychologists, not geneticists.

The Nazis used U.S. laws as a model in their own sterilization program (Chorover, 1979) beginning in 1934, which eventually sterilized over 200,000 people, mostly without consent and often without the individual's knowledge (Burleigh, 1995). The Germans were able to carry out such large numbers of sterilizations because they had the backing of an organized medical profession. The Nazis went further and exterminated hundreds of thousands of inhabitants of institutions for mental illness and mental retardation, using techniques that became a prototype for the gas chambers (Burleigh, 1994). Children were frequently starved to death on a special

diet. This program was designed to reduce the number of "useless eaters", not to affect the gene pool. The Nazis also rounded up families on registers for Huntington disease and exterminated them, in an attempt to eliminate HD entirely (Harper, 1992). Even the final eugenic attempt – extermination of Jews, Gypsies, homosexuals, and some Slavs – had no known effect on gene pools after killing 12 million people.

In recent years, it has come to light that many other nations besides the U.S. and Germany had eugenic sterilization laws. These nations include Austria, Brazil, Canada, Denmark, Finland, France, Norway, Sweden and Switzerland. Other nations with strong eugenics movements, such as the United Kingdom, never had such laws, preferring to rely on voluntary actions. In Latin America, eugenics developed largely as a theoretical movement not allied with medicine or human genetics (Stepan, 1991).

After World War II, U.S.- occupied Japan passed a Eugenic Protection Act (1948) allowing sterilization of persons with up to fourth-degree relatives with a list of presumably inherited conditions that looked remarkably like the lists in 1930's U.S. sterilization laws, but which omitted most major chromosomal and single-gene disorders. In most cases, sterilizations could only be conducted with the consent of, or at the request of, the individuals involved so, this was not a coercive eugenics law. The law limited abortions for "eugenic" reasons to conditions on the list. This meant that most abortions after prenatal diagnosis were done for "social" reasons (Ohkura 1989). The law was revised in 1996 to remove the word "eugenic" and the lists of conditions.

2.4.4 Eugenics in the World Today

There is little evidence for eugenics practice in the modern world, at least according to our definition as "a coercive policy intended to further a reproductive goal against the rights, freedoms, and choices of the individual".

Perhaps the best example was Singapore, which used monetary incentives to encourage reproduction among educated women and to encourage sterilization for uneducated poor women. China's law for Maternal and Infant Health Care (China, 1994) has aroused much attention, because it appears to require medical counselling before marriage for people whose families have a list of presumably inheritable conditions (including mental illness, epilepsy, feeblemindedness and other conditions listed in the old U.S. sterilization laws) followed by (if appropriate) sterilization or long-term contraception as a precondition of marriage. Another clause appears to require prenatal diagnosis for couples at risk, after which they should follow the doctor's advice. The law, however, carries no penalty and is not enforced. It appears to be closer to a "standard of care" than to a law, and the word "shall" may be better translated as "should" or "ought to", an ethical rather than a legal statement (Qiu, 1998). China's genetics profession, recognizing the importance of even a symbolic law, has requested change from the central government to bring the law into line with international standards of voluntary genetics services (Yang, 1998). Taiwan has had a similar law on the books for several years, without enforcement but also without arousing international attention. There appears to be little state-coerced eugenics in the world today. Nevertheless, we urge vigilance.

State-supported Programmes that are not Eugenics: Governments support many programmes, including some mandatory programmes, in the interests of public health, which do not constitute eugenics. These include:

- encouraging/discouraging births among the entire population. Although the WHO expert advisors reject coercive measures as restrictive of reproductive freedom, a government's attempt to control the quantity of its population is not eugenic as long as measures are used equally with regard to the entire population.
- laws prohibiting sex selection (India, China) are not intended to affect genetic characteristics.
- laws for the protection of the fetus from environmental harm. These may be described as "euthenic", or as part of general health care. As long as these do not coercively restrict the mother's activities, they would not be eugenic. An example is warning labels on alcohol and tobacco products about potential harms to the fetus, as mentioned above.
- laws for the protection and health of the newborn, including mandatory newborn screening for disorders where early diagnosis and treatment benefit the newborn.
- regulations establishing state-funded provision of genetics services, including genetic counselling, testing, prenatal vitamins (folic acid), prenatal diagnosis, special diets for mother or newborn. An example is the State of California provision of low-cost maternal serum alpha-fetoprotein testing in the United States. This programme is voluntary and has a refusal rate of 30% (Cunningham 1998). Although the medical setting itself establishes an uneven power balance between provider and patient, and the state-backed offer of services provides an incentive to accept these services, the programme is not intended to be coercive. Public health authorities in some nations, such as Denmark, require that physicians offer prenatal diagnosis to all pregnant women over age 35, but the woman has the choice to accept or decline the offer (Danish Council of Ethics, 1999). Care must be taken to ensure that people receive and understand full and unbiased information and that they understand that taking the initial blood test may lead to difficult decisions. Other examples include testing for spina bifida in the UK, and carrier and prenatal testing for beta-thalassaemia in Sardinia and Cyprus. In all three nations, testing is offered under the public health system, and affected births have decreased dramatically (Cuckle and Wald 1984; Cao et al 1989; Angastiniotis 1986). The programs were not, however, state coerced.
- laws regulating cousin marriages and other consanguineous unions. In some societies, these unions are preferred as a means of cementing social bonds. In some societies, the social and economic benefits of cousin marriage are regarded as outweighing the risk of having children with a recessive disorder (Jaber et al., 1998).
- regulations requiring addition of folic acid to cereal grains labeled "enriched". These are in the tradition of iodized salt or addition of vitamin D to milk.

"Quasi-eugenics": in Private or Community-based Programmes: These include testing required by religious communities as a precondition of marriage, or attempts by private groups to induce welfare mothers to be sterilized. Although these programmes are coercive, individuals may choose to leave a particular religious community or say no to a private offer. In a pluralistic society, communities may regulate the lives of their members, as long as individuals are not restrained from leaving the community. Private agencies are free to express their own beliefs.

"Economic Eugenics": Coercive eugenics tends to flourish in difficult economic times and this may be referred to us. Moreover, even in good times some social practices may approach eugenics even though, strictly speaking, they do not fall under our definition. These practices include non-availability or refusal of health care for fetuses with disabilities or their mothers, discrimination against prospective parents with disabilities that makes it difficult for them to reproduce, and discrimination against people with disabilities generally. The broadest discrimination and potential source of "eugenic effects" is against poor people generally in the health care system.

2.4.5 Threats to Freedom of Choice Exist

Free choice depends on 1) adequate and unbiased information; 2) availability of relevant alternatives, including care and therapy for children with genetic conditions, contraception, and legal abortion after prenatal diagnosis. For economic, religious, or political reasons, not all of these are available everywhere. In the absence of a full range of alternatives, accusations of "implicit eugenics" may be valid.

The most likely source of coercion in the modern world is biased counselling. Except in North America and English-speaking countries, much counselling appears to be either openly directive or provides intentionally biased information so that the persons counselled will do what the counsellor intends without the counsellor suggesting it directly (Carnevale et al 1997, Wertz 1998; Mao 1997). Much of this counselling is pessimistic. People have little recourse against biased information unless they are highly educated.

Another source of potential coercion is offering carrier tests during pregnancy rather than before conception. The medical setting tends to empower the provider and encourages such tests, especially among a vulnerable population, such as pregnant women. Pregnancy is not the optimum time to offer carrier testing for the first time.

New DNA chip technologies may reduce the pre-test counselling that can act as a safeguard against implicit coercion. With hundreds of tests in a future prenatal diagnosis, it will become impossible to describe, in counselling before the test, the details of each disorder for which a fetus will be tested. However, pre-test counselling should describe the general characteristics of the categories of disorders tested for (e.g., mental disability or neurological impairment). Women will receive intensive counselling after a fetal diagnosis.

2.4.6 Genetic Enhancement

Raising human capabilities into the "average" range may be regarded as treatment. Raising capabilities above the average is enhancement, historically called "positive eugenics". State-controlled programs requiring or encouraging reproduction among members of some social groups would be coercive positive eugenics, which is rare both historically and in the world today.

Individual choices favouring enhancement, while unlikely to change the gene pool unless practiced on a massive scale using germ-line gene therapy, have the power to alter society by creating classes of genetic "haves" and "have-nots". History teaches us that, if given a chance, we are likely to try to enhance ourselves. Human growth hormone, steroids, mood altering drugs, and special diets have all been popular attempts at enhancement. Gene therapy may be only one more addition to this already long list. In the increasingly autonomy-driven culture in much of today's world, individual demands may create a market for genetic enhancements, ironically leading to an autonomy-based individual "positive eugenics". For practical purposes, it may be impossible to limit or avoid these individual actions, even if a society wishes to do so.

Geneticists need keep in mind the ethical dangers of pursuing enhancement, including increased social inequality and a lowered tolerance for human diversity. Its consequences, intended and unintended, are not predictable at the present time. Enhancement would be 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 labeled as suffering is often the result of social conditions or social definitions of what is normal or desirable.

2.4.7 Genetic Determinism

The roots of all eugenics are racism and genetic determinism, the belief that individuals and populations can be equated with their genes, which determine their health, behaviour, and prospects in life.

2.4.8 Education, a Defense Against Eugenics

The best defense against eugenics is an educated public that knows how to ask for and obtain full and accurate information from health care providers and that does not hesitate to question the goals of testing and counselling. To this end, general science courses could include lessons on the history and outcome of past eugenic programs and also material on the fulfilling lives of many people with disabilities. Health care professionals could benefit from similar education. One way that professionals can learn more about the meaning of life with a disability is to work closely with patient and parent organizations. Improvement of services for persons with genetic disabilities should be pursued as a goal along with increased availability of counselling and prenatal choices.

2.4.9 Summary

In conclusion the word eugenics today 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 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 taking their chances of having an affected child, avoiding conceptions, using donor gametes, or using prenatal diagnosis followed by selective 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 (Angastiniotis et al 1986; 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 both the defects and the corresponding demand for 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 and other goals. This is the decisive difference between present day medical genetics and yesterday's eugenics.

2.5 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.

2.6 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.

3. Education as the Key to Ethical Genetics Services

3.1 Public Education

The goals of medical genetics can be optimally 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 individuals and families should participate in decision making (see 3.2 below). Users of genetic services 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 individuals and families be active participants, it is necessary that they receive some basic education about genetics.

In the long run, genetics education for the public can best be achieved through education in schools. In order that some genetics information can reach every pupil, it is important to define the fewest, simplest and most up-to-date pieces of information that can be included in biology teaching in schools and to define the most appropriate stage for teaching them. The scientific community in universities must take greater responsibility in the reform of school curricula, and in working with school teachers to revise school science texts and formulate an ideal science curriculum for each stage that addresses basic genetic concepts and principles, including simple facts on specific local problems and how to deal with them.

Education of the public outside schools depends to some extent on education of the media and education of health workers in contact with individuals at the primary health care level. Combining educational goals with community genetic services and prevention measures

integrated into primary health care may prove very valuable in disseminating the correct genetic information to the population in general.

Genetics centres may 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 the needs of individuals and families. In a team approach, professionals from different specialities will feel free to call upon each other to provide areas of expertise with which they are not explicitly familiar. A physician or genetic counsellor may wish to call upon a social worker to inform individuals and families 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 lay people and professionals is generally best if the professional is familiar with the individual's or family'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. The objectives of medical genetics education may be fulfilled if students can acquire the following:

- Sound basic knowledge of genetic mechanisms in health and disease, an understanding of new genetic technologies and their application in medicine for diagnosis, prevention and treatment.
- An understanding of the basic principles, ethics and approaches for genetic counselling.
- Knowledge of the genetic services available in the community and how to refer people who need more specialized genetic services.
- Knowledge of the common genetic problems in the community and the strategies for management and prevention.
- Knowledge about human diversity and variability.
- An attitude of life-long education and self-education which is necessary in the rapidly expanding field of genetics.

3.2.2. Ph.D. Medical Geneticists

A sizeable number of medical geneticists are Ph.Ds in biology, who may provide genetic analysis and genetic counselling. They should receive training in counselling and ethics.

3.2.3 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.

3.2.4 Midwives

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

3.2.5 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 the needs of individuals and families because of its emphasis on counselling. Counsellors' training is less lengthy and less costly than training physicians. Training programs in genetic counselling should be encouraged in all nations, regardless of whether genetic counselling exists as a separate profession.

3.2.6 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 anaemia in the USA.

3.2.7 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.

3.2.8 Community Health Workers

These would also include primary health workers, rural health workers and midwives. They are directly in contact with the largest portion of the population within the health sector. Their proper education and training related to basic genetics offers considerable potential benefits for prevention of genetic disorders. Training courses could include education in family history-taking, basic training in counselling skills, detection of high-risk families, guidelines in delivering pre-conceptional counselling for the prevention of genetic and congenital disorders, and guidelines on prenatal and postnatal services.

3.2.9 Pharmacists

Genetic conditions may affect responses to drugs, and genetic variation in several enzymes relevant to drug metabolism are known. Pharmacogenomics is rapidly developing, and it is important that pharmacists as well as pharmacologists are aware of differing responses to drugs. It is predicted that genotyping of relevant enzyme loci will become routine prior to drug treatment. This is an area where genetics will change medicine.

3.2.10 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.

3.2.11 Oral Health Professionals

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

3.2.12 Laboratory Personnel

Laboratory personnel, whatever their education, should be made aware of the ethical responsibilities connected with their work and with the possibility that unexpected results of analyses could reflect genetic variation.

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 information about individuals and families and keep records. These personnel should not be regarded as merely peripheral to patient care. In some cases, such personnel will have extensive on-going contact with individuals and families and may even, perhaps inadvertently, make decisions that affect these individuals and families' health. It is important that such persons receive some education about common genetic disorders so that they can better communicate with individuals and families. 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. Clergy are most often consulted as to the ethical aspects of health and reproductive choices. Some basic human biology, with emphasis on genetics and bioethics, should be included in their standard curriculum and continuing education.

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 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 our offspring or we 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, pre-conceptional care, prenatal care, paediatric 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).

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 culturally and legally acceptable reproductive alternatives with both parties present. Geneticists need to 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. Carrier-carrier couples or persons with genes for X-linked or autosomal dominant disorders obviously have the right to marry and make their own choices.

4.2 Family Planning

Genetics services should be included in larger family planning programmes that present couples with the full range of options described under below (4.3.4), including full information about contraception.

4.3 Preconception Counselling

4.3.1 Family History

Couples intending to have children should be encouraged to meet with their physician or a professional with training in medical genetics 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.

4.3.2 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.

4.3.3 Counselling High-Risk Couples

In cases where a couple's chance of having a child with a genetic condition 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.

Chances of having children with genetic conditions or chromosomal abnormalities associated with advanced maternal and paternal age should be explained. Couples may be encouraged to complete their childbearing before the ages of highest risk if the alternatives of prenatal diagnosis and termination of pregnancies with an affected fetus 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.

4.3.4 Assisted Reproduction and Medical Genetics

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

4.3.5 Children's Rights to Medical Information

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 voluntary consensual registers of donors or birth parents to whom disclosure is acceptable, including records of the genetic health history of the biological father in cases of non-paternity. These registries should be periodically updated and registrants should have the option of removing their names. Anonymity must be guaranteed, unless both parties agree to disclosure of names. On reaching the age of legal majority, adopted children should, if possible, be provided with a genetic health history of their biological parents, if they wish it, even if names are not revealed.

4.3.6 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 the desire to be a responsible parent and help prospective parents with disabilities to evaluate their abilities to care for a child. 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 who would have a reasonable expectation of leading a healthy life. In this context, a criterion for counselling is to help parents with disabilities evaluate how an affected child's prospects in life would compare with those of other children in the child's birth cohort. Also, persons with disabilities, including blindness and deafness, should not be excluded from adopting children, provided that they can care for a child.

4.4 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:

4.4.1 Preconception Risk Assessment

Preconception risk assessment offers the opportunity to identify:

- Individual and social conditions, e.g., extreme obesity; advanced maternal age; special diets; 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.

4.4.2 Preconception 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.

4.4.3 Preconception Visit

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.

4.4.4 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.

4.5 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 of recessive conditions or one partner has the gene for a dominant condition, or the wife has the gene for an X-linked condition. 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.

4.6 Childhood Care

Today most persons brought into contact with a genetics clinic are children under the age of 14. The health care professional 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 professional should regard them equally with the parents of "normal" children. The professional 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 reaches the father, lest he blames 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 professional 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.

4.7 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 speciality related to paediatrics or obstetrics to an adult speciality closely allied to general practice and adult type preventive medicine. Genetic risk testing could become part of routine physical examinations. Genetic testing prior to drug treatment to tailor drug therapy to the individual will be a major activity in the field of medical genetics.

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. Each country will set its own priorities in health care according to the country's laws, tradition, and culture.

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, treatment, and other legally and culturally accepted services should be reimbursed by 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. All services, except newborn screening for treatable diseases should be on an individual and voluntary basis. As a rule, fees for services might be requested, but reduced or waived in proportion to an individual's or family's inability to pay.

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. Selective abortion of a wanted 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 whom 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).

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

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

Part II
Specific services

1. Basic Principles in Interactions between Professionals and Laypersons

Both medical geneticists and persons receiving counselling have responsibilities.

1.1 Respect for Persons

Respect for persons should be the basis of all genetics services (Tables 2 and 3). This is not only the most ethical approach, but also the most effective in terms of communication and care. Geneticists should regard individuals and families as partners in their own care. This means respecting people's intelligence, whatever their level of education. It means listening to people 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). People's 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 person's level of current understanding. The approach avoids making already knowledgeable people 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 individuals and families need emotional support. The supportive aspect of counselling is of at least equal importance with the informational aspect. The counsellor's presentation and demeanour should convey acceptance of those receiving counselling 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 need to consider the integrity of the entire family, even if only one member comes for counselling. In Asia particularly, harmony or concordance is one of the most respected principles in maintaining the family. It is essential that the family remain at peace, with caring decision-making and a balance of power among family members.

2. Genetic Counselling

The ethical principles in Table 1 also underlie genetic counselling (Table 3).

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 3).

Table 3. Ethical Principles Applied to Genetic Counselling

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

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

Optimum reproductive 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.

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 M.D., Ph.D., nurse, social worker, or specially trained genetic counsellor.

It is recommended to have a preliminary session for blood sampling and planning of information collection as well as to prepare people, in advance, about what they should expect from the main counselling sessions. 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 Special Aspects of Communication in Asia

In Asia, especially, much communication is implicit rather than explicit. In general, people do not like to discuss some problems openly. Traditionally, to speak less is better than to speak too much, as "Silence is Golden". Therefore, people coming to counselling may not open their minds or hearts to the counsellor or explain what is causing their hidden distress. The counsellor's role is to give relief from fear or anxiety, by paying attention to emotional distress and bestowing confidence.

2.1.2 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 individual's or family's group are of paramount importance. The counsellor need not be from the same group as the persons receiving counselling, though in some instances this may be desirable. There are both advantages and disadvantages of having counsellors from the same cultural or ethnic group as those being counselled. On the one hand, a counsellor from the same group may have first-hand knowledge of the values, beliefs, and practices prevalent in the individual's or family's social environment. On the other hand, even when the counsellor and persons receiving counselling 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 persons from their own communities than they are with people from other communities (Fisher, 1996).

Ideally, the counsellor should speak the language of those being counselled. If an interpreter must be used, the interpreter should have special training in communicating with persons in medical settings, and should also have some basic knowledge about medical genetics.

2.1.3 Non-Directiveness

Counselling should be non-directive, except when treatment is available. Non-directive counselling is the provision of accurate, full, and unbiased information in an empathic relationship that offers guidance and helps people work through to their own decisions.

Non-directive counselling has two major elements. The first is provision of accurate, full, and unbiased information that individuals and families may use in making decisions. The second is

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

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

Non-directiveness does not mean that the counsellor should be without a set of values. Most people prefer to think that they are interacting with a morally concerned human being rather than a 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 individuals or families, 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 people. 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 people can make informed decisions: and (b) tell people 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 people 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 individual or family a referral.

If asked, it may be appropriate to tell people 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 options. Counsellors should be aware that families and organizations for people with genetic disorders could present biased views.

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

history, and situation. The counsellor should make clear that the individual or family must make her/his/their own decision and that the counsellor's choice of action may be irrelevant to anyone else'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 may also refuse to perform or to offer referrals for non-medical services such as sex selection. Such refusals do not contravene the ideals of non-directive counselling.

2.1.4 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 people 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 also 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 individual or family 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.

2.1.5 Presentation of Risk

When risks are involved, whether these be chances of having a child with a genetic condition or risks arising from a procedure or treatment, counsellors should present these chances or 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 chances. There is a tendency for people to regard a proportional risk as higher than the same risk given as a percent. Hence the importance of presenting a chance or risk both ways. Some people may tend to overestimate small chances or to underestimate high chances (as long as these do not reach 100%). Many people tend to interpret the same numeric risks as lower than do counsellors (Wertz et al, 1986). People also tend to interpret chances in binary ("either/or") form, whatever the level. Counsellors should not expect people 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 people to make decisions, counsellors should ask them to envisage the future consequences of each alternative, considered over the life course. People should be asked to consider the consequences of each choice for themselves, their spouse or partner, family, and children.

2.1.6 Contexts of Counselling: Settings and Scheduling

Persons receiving unfavourable diagnostic test results (prenatal, paediatric, 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 people 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 users of genetic services travel. These centres should be available to public transportation. Appointments should be scheduled with consideration for the individual's or family'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 people should be served promptly on arrival at the clinic. If a waiting period is usually necessary after arrival at the clinic, people 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 persons receiving counselling must be able to meet in a private room with the door closed. Childcare 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, people should also have the opportunity to discuss the test(s) with a knowledgeable person (perhaps a community health worker) before testing.

2.1.7 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, 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, because most people will find these terms confusing. The term "family history" is preferable to terms employing the word "pedigree", which some people associate with animals only.

2.1.8 Counselling Spouses/Partners

If a couple intends to have children, both partners should usually be offered counselling 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.

2.1.9 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 those from prenatal diagnoses. Follow-ups should be voluntary, however.

2.1.10 Summary of Counselling Session Provided

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 individual or family to summarize the session briefly from their point of view. The purposes of this final summarizing are (1) to refresh the individual's or family's memory; (2) to help the counsellor evaluate the individual's or family's level of understanding of medical/genetic knowledge; and, (3) to help the counsellor evaluate the individual's or family's need for further supportive counselling or referrals.

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

2.1.11 Materials Provided to People Receiving Counselling

If appropriate, counsellors should provide educational materials appropriate to the individual's or family's level of literacy. If someone 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 persons will desire information beyond that usually presented in informational brochures. Some people will wish a list of publications available at their libraries, and some will wish to look at the medical literature itself or sites on the Internet. Counsellors should be able to provide up-to-date lists of publications at different levels.

2.1.12 Evaluation of Counselling

Those providing counselling should have evaluation measures in place to assess the quality of communication, understanding of information, and usefulness of counselling to people's 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 people 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 people's understanding; and, (2) people's reports (or other evidence) that counselling assisted them to make decisions that were best for them, 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 individual's or family's permission) and criticizing each other's work in a non-judgmental manner; and, (3) individual or family review, using periodic surveys or interviews.

2.2 Counselling Children and Adolescents

2.2.1 Involving Children in Decisions About Testing and Treatment

Whenever possible, professionals should promote understanding in children and adolescents about their disorder and alternatives for treatment. Aspects of the disorder and possible treatment should be discussed with the parents, in the child's presence. Parents should make decisions regarding therapy or preventive measures. Children should not be tested for disorders of later onset in the absence of treatment 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 an 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 an adolescent to contribute to a decision about testing, treatment or prevention 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 individuals or families) is a commitment of genetics professionals. This assumes that

all relevant facts are known to those receiving counselling, and efforts are made to encourage people to consider the facts in the context of their 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. Some persons counselled may be mentally ill, severely retarded, or abusers of alcohol or drugs. Some people may be severely disadvantaged in communication because of poor education, although they are of normal intelligence. For these reasons such persons 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 persons or to impaired individuals themselves is ethically acceptable, in exceptional cases, if the likelihood of harm to others is great and if the geneticist has informed the individual 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 layperson voluntarily assigns the power of decision to an expert. This model should be used only as a last resort and only if the individual insists on it.

2.5 Effects of Professionals' Gender

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

3. Rights to Referral

If a professional is unable or unwilling to perform a medically indicated service for personal moral reasons, the professional is obligated to refer the individual or family 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. This does not mean that professionals should offer referrals for all requested services. A professional need not refer for sex selection in the absence of an 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 professional has a moral duty not to abandon women or families 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, physicians who perform prenatal diagnosis owe the woman help and support for her choices after receiving results.

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

4. Duty to Recontact

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

Genetics professionals have an ongoing duty, unless instructed otherwise by the individuals or families, to inform them about new tests and treatments. In genetics, the ethical responsibility to follow up and recontact families may extend for several generations. The professional's ethical duty extends beyond those individuals who have presented themselves for care. 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 offer individuals and relatives the opportunity 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 8.2.8 below, such registers would cause harm to individuals and families only if data protection is not strict. Mechanisms for ensuring privacy must be established.

5. Screening and Testing

5.1 Definitions and Requirements for Programmes

5.1.1 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 haemoglobin disorders. 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.

5.1.2 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.

5.2 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.

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.

5.3 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 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.

The introduction of multiplex screens such as tandem mass spectroscopy raises new ethical issues, because it may lead to the identification of diseases that are not treatable at the present time. There are both benefits and risks associated with knowing that an apparently healthy newborn will develop one of these diseases early in life. For some parents, the knowledge may lessen self-blame and prevent weeks or months of searching for a diagnosis. Parental knowledge may also confer a benefit to the child, because parents could be prepared to take advantage of new and rapidly-evolving treatment. So, on the other hand, some parents may not wish to know, preferring to enjoy the months or years before symptoms appear. On balance, it appears that the benefits of parental knowledge outweigh the risks. However, parents who do not wish to know about currently untreatable disorders should have the opportunity to “opt-out” from receiving this information.

The uses of multiplex screens (if any) will necessarily vary according to country. Screening for treatable disorders, where early treatment is effective, should have priority over identification of other disorders.

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 purpose of informing parents about the newborn's carrier status is for the benefit of the parents' own reproductive plans. 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. Geneticists should weigh the 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.

5.4 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).

5.5 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, pilots, 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.

5.6 Premarital Screening for Carrier Status

Premarital screening for carrier status for disorders common in a community 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.

5.7 Summary: Ethical Aspects of Genetic Screening and Testing

Suggested ethical guidelines for screening and testing are listed in Table 4:

Table 4. Proposed Ethical Guidelines for Genetic Screening and Testing

1. Genetic screening and testing should be voluntary (autonomy); not mandatory, with the exception noted in the last point below.
2. Genetic screening and testing should be preceded by adequate information about the purpose and possible outcomes of the screen or test and potential choices to be made (autonomy, non-maleficence).
3. Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened (autonomy).
4. Results should not be disclosed to employers, insurers, or others without the individual's consent, in order to avoid possible discrimination (autonomy, non-maleficence).
5. In rare cases where disclosure may be in the best interests of the individual or of public safety, the health provider may work with the individual towards a decision by him or her (beneficence, non-maleficence, justice).
6. Test results should be followed by genetic counselling, particularly when they indicate the presence of a mutation or a genetic condition (autonomy, beneficence).
7. If treatment or prevention exists or is available, this should be offered with a minimum of delay (beneficence, non-maleficence).
8. Newborn screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn (beneficence, justice).

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

Genetic testing is the analysis of the status of a particular gene. A genetic test may establish: (a) a specific diagnosis of a genetic condition in a symptomatic individual, b) the certainty that a particular condition will develop in an individual who is asymptomatic at the time of the testing

(presymptomatic diagnosis), or c) the presence of a genetic predisposition to develop a particular complex disease such as cancer or cardiovascular disease (predictive genetic testing). The main objective of genetic screening and testing is to prevent disease or secure early diagnosis and treatment.

Ordinarily, population-screening programmes are offered only when proven methods of treatment or prevention are available. In selecting population groups to be screened, it is important to avoid the possibility of stigmatizing the entire group. Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened in the absence of preventive or therapeutic options for the individuals screened.

Screening programmes are usually better received if they work in cooperation with community leaders in the group to be screened. Educational programmes for the group should precede screening.

If screening is provided for newborns, there is an obligation on health care providers to make sure that appropriate and timely treatment or preventive measures are available.

6. 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 person 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 person consents voluntarily. The elements of informed consent appear in Table 5 overleaf. The purpose of informed consent is to make certain that people 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 non-paternity as an incidental finding, this also should be included in the description of risks. If results may be ambiguous, people 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 persons 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 individual’s or family’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.

Table 5. Autonomy and Informed Consent

a) Applicable to clinical practice:

Genetic testing in clinical practice should be voluntary and should occur in the context of a comprehensive genetic service and valid process of informed consent, with an explanation of the following elements:

- The purpose of the test.
- The chance that it will give a correct prediction.
- The implications of the test results for the individual and family.
- The tested person's options and alternatives.
- The test's potential benefits and risks, including social and psychological.
- That social risks include discrimination by insurers and employers (even though this may be illegal); and
- That whatever decision individuals and families make, their care will not be jeopardized.

b) Applicable to Research and Quality Control:

Elements of a valid informed consent process include an explanation of:

- The experimental nature and purpose of the study.
- Why the individual is invited to participate and that participation is voluntary.
- A description of the procedure.
- The discomforts and risks (if any) of the test to both the individual and the family.
- The uncertainty of the results of the test for prediction and accurate genetic counselling.
- The possible benefits to others and to science.
- The confidentiality of records identifying the tested individual.
- Whom to contact for questions about research or in the event of a research injury.
- The right of the individual to withdraw at any time, and
- The right of the individual and family to unrestricted health care, even if the individual withdraws.

Genetics professionals should attempt to evaluate understanding, especially for procedures involving higher risks. One way to evaluate people's 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 person 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 individual wishes to go ahead. To withhold a non-experimental procedure because a competent person 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.

7. Presymptomatic and Susceptibility Testing

7.1 Definitions

Presymptomatic testing (e.g., for Huntington disease) identifies individuals who will develop a genetic disorder if they live long enough. Susceptibility testing (often referred to as 'predictive 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.

7.2 Benefits and Risks

7.2.1 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.

7.2.2 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.

7.3 Recommendations for Offering Tests for Susceptibility to Common Diseases

Testing of individuals with a family history of disease:. 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.

Privacy: 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.

7.4 Recommendations for Offering Presymptomatic Tests

Presymptomatic testing should be available for adults who want it, even if there is no prevention or treatment for the disorder, 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 probabilities that are not close to 0% or 100%, 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. (For treatable or preventable disorders, testing may be carried out in the interests of the person’s health.)
- There is evidence that the information provided by testing would be used to prevent harm to the tested individual, spouse, family, prospective children, or others.
- Testing is accompanied by a counselling programme appropriate for the disorder. 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, adjusted to the needs of individuals, is preferable to a protocol specifying a certain number of sessions. If married, individuals should be offered counselling together with their spouses for some of the sessions. This is especially important if children are contemplated.

7.5 Summary of Presymptomatic and Susceptibility Testing

There are important differences between presymptomatic and susceptibility testing. Presymptomatic testing refers to identification of healthy individuals who may have inherited a gene for a late-onset disease, and if so will develop the disorder if they live long enough (e.g., Huntington disease). Susceptibility testing identifies healthy individuals who may have inherited a genetic predisposition that puts them at increased risk of developing a multifactorial disease, such as heart disease, Alzheimer disease or cancer, but who, even so, may never develop the disease in question. Proposed guidelines for presymptomatic and susceptibility testing are presented in Table 6.

Table 6. Proposed Guidelines for Presymptomatic and Susceptibility Testing

1. Genetic susceptibility testing of persons with a family history of heart disease, cancer, or other common diseases of possible genetic origin should be encouraged, provided that information from the test can be used effectively for prevention or treatment (beneficence).

2. All susceptibility testing should be voluntary, preceded by adequate information and based on informed consent (autonomy).

3. Presymptomatic testing should be available for adults at risk who want it, even in the absence of treatment, after proper counselling and informed consent (autonomy).

4. Testing of children or adolescents should be carried out only if there are potential medical benefits to the child or adolescent or if an adolescent requests it for purposes of reproductive decision-making (autonomy, beneficence).

5. Employers, insurers, schools, government agencies or other institutional third parties should not be given access to test results (non-maleficence).

Presymptomatic testing in the absence of therapeutic options should be available if the following conditions are met:

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

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

8. 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 an individual or family 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.

8.1 Preparing People before Testing

In general, it is best to prepare individuals 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 may be warned of this possibility before testing. In nations where this is possible, the mother could be counselled individually before a couple is seen together, so that she can withdraw from testing if non-paternity 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 needs to 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.

People 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. Individuals should be informed in advance of the clinic's policy on disclosure to relatives at genetic risk (see 8.2.2 below), and relevant laws or regulations.

8.2 Situations Involving Disclosure and Confidentiality

8.2.1 Full Disclosure of Clinically Relevant Information

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 person and allows the person to make decisions. Counsellors need to be aware, however, of the potential emotional impact of disclosure in societies where hereditary disorders are associated with shame or guilt.

Although people may have a "right not to know" genetic information, the right not to know presumes that people 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. 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 relationship between health care provider and layperson. If the health care provider edits vital facts out of the communication, the relationship is less than optimal and can be harmed. If the individual or family later discovers non-disclosure, confidence in health care providers could well

be shaken or undermined and result in further harm. People 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 an individual 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 person 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 would 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 in cultural context: in some cultures, particularly in Asia, any disclosure of a hereditary disorder in a family may be considered shameful. Even if only the affected individual is told, knowing about the disorder means losing face and dishonoring the family name. Parents, especially mothers, feel guilty. Most parents will not be open with counsellors and may wish to keep their child's condition secret. Full disclosure of information may have the effect of alienating spouses or members of both extended families. Counsellors need to consider the effects of full disclosure on all concerned and to manage the disclosure carefully.

Disclosure of normal test results: individuals 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., non-paternity, fetal sex in the absence of X-linked disorders) may be withheld if this appears necessary to protect a vulnerable party. People 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, national law regulates handling of non-medical results.

Prior disclosure to another party: sometimes an individual 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 individual her/himself in a timely fashion.

Selective non-disclosure at individual's request: people 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 person 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 person 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 person's best interests and should not be encouraged. People 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.

8.2.2 Confidentiality when other Family Members are at High Risk

In genetics, the "true patient" may be 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 individuals 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 individual or parents help in contacting relatives so that they may be informed about specific risks. The first contact with the individual 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 individual'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 the individual'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 people of these obligations. "Non-directive counselling" is not appropriate in these situations. Usually individuals will cooperate if repeatedly urged to do so.

Sometimes an individual prefers that the professional, rather than the individual, tell relatives. The individual may feel embarrassed about transmitting bad news, but also thinks that the relatives should know. The professional should offer people the option of having the professional tell the relatives, at the person'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 individual's request.

In asking people 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 individual's rights to confidentiality. In some cases, as when a family feud exists, the individual 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 professional 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, professionals 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 to prevent harm.

The professional may attempt to warn relatives who are at high risk of serious harm, even against the individual's wishes, provided that the four conditions described below are met (and that the warning can be done without unduly disturbing the basic harmony in family relationships). The professional 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 professional should take care not to identify or describe the genetic status of the original individual, except with the individual's permission.

In rare cases an individual refuses to disclose or to permit disclosure of information to relatives at genetic risk. According to the U.S. President's Commission, a multidisciplinary body of ethicists, clinicians, and legal experts, the genetics professional may, unless it is prohibited by law, override individual confidentiality if the following four conditions are met (United States, 1983). The Institute of Medicine (1994) and the American Society of Human Genetics (1998) supported these recommendations.:

- 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 individual 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, but not required, 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 required to disclose information about relatives' own genetic risks if the relatives ask. In this case the relatives have become patients, asking for information about themselves. Professionals should not be legally required to disclose such information to relatives who do not ask.

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, at the invitation of the original individual counselled, 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 individual. Family members may, of course, learn the individual's diagnosis indirectly as a result of their own counselling and tests. This is probably impossible to prevent. Nevertheless, the individual should not be able to prevent others from learning genetic information about themselves.

Cases of outright refusal to contact relatives will, however, continue to occur. If geneticists have informed people at the outset about the need and duty to inform other family members who have a reproductive or health risk, and have also informed them that confidentiality is limited by this moral duty, medical geneticists have laid the groundwork for action if the individual 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 person 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 may be ill advised to permit laypeople 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 individual to keep the risk a secret. Professionals can make it clear to an individual that if the individual will not carry out his or her own duty, this refusal place the professional in an intolerable position. If a history of alienation and emotional problems in the family emerges, the individual can be offered help from a mental health specialist with the task of disclosure.

One professional society has recommended that an ethical review committee should make the final judgement about whether relatives should be contacted in individual cases. Guidelines issued by the Japan Society of Human Genetics in September 1995 refer these disclosure dilemmas to ethics committees. The Japanese guidelines state that "if the sharing of information with another specific person (family member at present or in the future) will avoid serious injury to that person, it is necessary to seek the consent of the subject to reveal that information, and even if agreement cannot be obtained, if it is judged necessary the obligation of confidentiality can be broken. Such an exception must be made following the judgement of the responsible ethics committee, not by the counsellor. Therefore, in our opinion, in such cases, the committee, not a single counsellor, will decide if disclosure of the information to the relatives should be made" (Japan Society of Human Genetics, 1995).

This approach has both advantages and disadvantages. It appears that for disorders that are treatable, such as familial polyposis coli, familial hypercholesterolaemia, and some cases of breast cancer, individual geneticists or genetics centres could make the decision without ethical review. However, there could be a temptation on the part of geneticists to include a wide variety of disorders under the heading "treatable", even if treatments are not especially effective, leading to the danger of unwarranted breaches of confidentiality. There are also disorders that are not treatable but could be prevented in future generations if prospective parents become aware in time. Placing the burden of weighing benefits against harms of disclosure in these situations on individual geneticists could be unfair to the geneticist. Using an ethics committee as an intermediary may be an effective solution, especially if the geneticist is facing a problem where

he or she would like a second opinion. This may also be provided through a system of ethical consultations.

8.2.3 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 professional 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.

8.2.4 Spouses/Partners

Confidentiality may 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 have a moral obligation to share information with their partners in order that both are aware of potential harms to a future child. Professionals have a moral obligation to remind people of this. In some rare cases, professionals may be permitted to tell spouses/partners without the individual'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 8.2.2).
- An individual’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 non-paternity after a test taken for another purpose. The information is primarily the individual’s and the individual 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 individual 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 professional may support a decision not to disclose.

- Non-paternity: ideally, the counsellor may be able to prevent the situation of disclosure of an incidental finding of non-paternity by telling the woman alone, before testing, that the test could reveal non-paternity. 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 non-paternity.

Professionals 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 professional 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 non-paternity. There is rarely a justification for a professional to reveal incidental findings of non-paternity 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 non-paternity 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.

8.2.5 Non-Biological Relationships other than Non-paternity

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 needs to weigh carefully the possible benefits and harms of disclosure; disclosure may do more harm than good, unless required to prevent serious genetic harm to living or future persons.

8.2.6 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 private 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. 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 might 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, private health 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 genetics professionals around the world agree that employers, insurers, and other institutions, such as schools, should not have access to an individual's test results without the individual's consent; (Wertz and Fletcher, 1989). This applies to carrier tests, presymptomatic tests, and susceptibility tests. Information about someone who is symptomatic for a disease may be conveyed to institutions as part of general medical information. Thus, information about sickle cell carrier status would be withheld, while information about the medical status of someone with sickle cell disease would be disclosed to a school or employer who requested it.

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 - there is a near-universal right to a minimum amount of life insurance, regardless of risk.

There are two basic approaches to protecting individuals in the areas of employment or private health insurance. 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. According to this view, employers and insurers should be prohibited by law from requiring presymptomatic tests or susceptibility tests as a condition for employment or insurance, 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 and insurance 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, insurers or prospective employers should have no access to the child's test results. Increasingly, discussions of genetics and insurance involve whether or not to include family history – a cornerstone of insurance practice – under the heading of "genetic testing and genetic information". Although inclusion appears logical, it could substantially change insurance practice.

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.

8.2.7 Other Institutions

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

8.2.8 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. However, registers have been used in government coerced eugenics programs in the past (Harper 1992). Therefore the utmost caution is necessary. 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.

8.3 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 need to be aware that medical records, including family histories, typically pass through many hands. Information not 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 a person’s uncle spent time in jail. 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. People receiving genetic services should have the opportunity to see or hear all information about them that will be referred to other professionals. 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, 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. Ideally, 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 individuals or relatives should not be provided to third parties without the person’s explicit request or consent, and should not be provided to commercial entities at all.

8.4 Summary on disclosure and confidentiality

Disclosure and confidentiality issues are some of the most frequent ethical problems appearing in medical genetics. Because of the possibility of harm from disclosure to institutional third parties, utmost care must be taken to protect confidentiality. Suggested guidelines are summarized in Table 7.

Table 7. Disclosure and Confidentiality

1. Professionals should disclose to tested individuals all test results relevant to their health or the health of a fetus. Adequate information is a prerequisite for free choice and is necessary to the open communication and trust that should mark the relationship between the provider and the person counselled.

2. Test results, including normal results, should be communicated to the tested person without undue delay.

3. Test results not directly relevant to health, such as non-paternity, or the sex of the fetus in the absence of an X-linked disorder, may be withheld if this appears necessary to protect a vulnerable party or if prescribed by national law.

4. The wishes of individuals and families not to know genetic information, including test results, should be respected, except in testing of newborn babies or children for treatable conditions.

5. Information that could cause grave psychological or social harm may be temporarily withheld. Within the general duty of disclosure, the counsellor may exercise judgement about when a tested person is ready to receive information.

6. If a couple intends to have children, individuals should be encouraged to share genetic information with their partners.

7. Where appropriate, as part of their general duty to educate, counsellors should inform people that genetic information may be useful to their relatives and may invite individuals to ask the relatives to seek genetic counselling.

8. The provision of genetic information to relatives about the family so as to learn their own genetic risks should be possible, especially when harm can be avoided.

9. Results of carrier tests, presymptomatic tests, susceptibility tests, and prenatal tests should be kept confidential from employers, health insurers, schools and government agencies. People should not be penalized or rewarded for their genetic constitutions. Information about a symptomatic condition may be disclosed as part of general medical information, in accordance with laws and practices in different countries.

10. Registries (if any) should be protected by the strictest standards of confidentiality.

9. Testing Children and Adolescents

9.1 Guidelines for Testing

The following suggested 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 not before this time.

- The test has no health benefits for the minor, but may be useful to the minor in making reproductive decisions in due time. 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). Some parents' groups have supported carrier testing on the basis that if the family is sufficiently educated about genetics, there is a minimal possibility of harm and both child and parents may benefit from knowing before the child reaches the teenage years (Genetic Interest Group 1996). Such requests should be evaluated on an individual, case-by-case basis. For presymptomatic, (e.g. Huntington's disease) or susceptibility testing (e.g. cancer mutations), harms would appear to outweigh any potential benefits. Parents' groups have opposed such testing (Genetic Interest Group, 1996).
- 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 usually best avoided. It is not "necessary" medical care and does not relate to reproductive rights.

  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 and 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 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.

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.

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 professionals should not yield to their request.

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 an object undergoing quality control (see Adoption 11 below). However, a family history of the birth parents should be provided, if possible, just as a family history is usually available for biological children.

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 ethical committee, established by the clinic for this purpose, would help to mediate disputes within families. Alternatively, the conflict could be handled in an ethical consultation.

9.4 Disclosure of Test Results to Children

It should not be assumed that parents would 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; Institute of Medicine, 1994). Scientists should avoid presenting findings to the media that could lead to premature genetic explanations for common behaviours (e.g., violent crime, and 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, in so far as possible within a country's culture and tradition. 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 9.1). Children should not be tested for later-onset disorders before adoption (American Society of Human Genetics, 2000). 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 find homes for 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 be able to learn, if they wish, 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.

Keeping in mind the past misuses of registers (Harper 1992), private agencies or nations could 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 may be notified of the causes of their biological parents' deaths, if they wish to know. Persons who were adopted or who placed a child for adoption in earlier years may be informed of the existence of such registers and should be given an opportunity to enter information if they wish. The registers may include information about the health of siblings and half-siblings. Before establishing a register, the parties concerned should be certain that it can be protected from misuse.

Such registers may also be used for transmitting names of biological parents and children, but only if both parties enter into the record a statement of 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 or isolation of fetal genetic material 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 a couple important information about the fetus. It may also help the professional team to prepare for a difficult delivery. Ethical guidelines for the provision for prenatal diagnosis are given in Table 8.

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. In some cases, information from prenatal diagnosis may make delivery safer for both the mother and child, for example by planning to have the birth in a hospital with special facilities for high-risk births. Prenatal diagnosis of lethal conditions such as anencephaly could avoid unnecessary obstetrical procedures in the mother. The majority of medical geneticists regard these as medically indicated uses of prenatal diagnosis. Prenatal diagnosis should be offered to all pregnant women at elevated risk, regardless of their views on abortion. It is unfair to withhold information about 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.

Table 8. Proposed 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, or any other considerations (justice).

2. Prenatal diagnosis should be voluntary in nature. The prospective parents should decide whether a genetic disorder warrants prenatal diagnosis or termination of a pregnancy with an affected fetus (autonomy).

3. If prenatal diagnosis is medically indicated, it should be available regardless of a couple's stated views on abortion. Prenatal diagnosis may, in some cases, be used to prepare for the birth of a child with a disorder (autonomy). Prenatal diagnosis for adult-onset disorders may require special counselling , so as to avoid testing of children who may be carried to term.

4. Prenatal diagnosis is done only to give parents and physicians information about the health of the fetus. The use of prenatal diagnosis for gender selection, apart from a situation with risk for sex-linked disorders, is not acceptable (non-maleficence). Prenatal diagnosis for paternity testing, except in cases of rape or incest, is generally unacceptable, but should be considered on a case-by-case basis.

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 (justice).

6. Counselling should precede prenatal diagnosis (non-maleficence).

7. Physicians should disclose all clinically relevant findings to the woman or couple, including the full range of variability in the manifestations of the condition under discussion (autonomy).

8. The woman's and/or couple's choices in a pregnancy with an affected fetus should be respected and protected, within the framework of the family and of the laws, culture and social structure of the country. The couple, not the health professional, should make the choice (autonomy).

Most women who request prenatal diagnosis in order to "prepare themselves for the birth of a child with a disorder" hope for favourable 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 of the prenatal diagnostic procedure 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 that their child will have a genetic condition to make early plans for treatment, housing, and education. Some couples change their minds about abortion after receiving results indicating the presence of a potential disability. To refuse to offer prenatal diagnosis is to prejudge a couple's behaviour. In fact it may be difficult to predict responses to a particular test result.

Prenatal testing for disorders that appear in adulthood, such as Huntington disease, poses difficult ethical problems. If parents are adamantly opposed to abortion, the information from the test provides no benefit to them or to the child, and may cause substantial harm to the child after birth, from stigmatization by family and society. If, after counselling, parents are still unwilling even to consider abortion, yet desire the information, the most ethical course of action may be to withhold the test. Performing a test for an adult-onset disease on a fetus that will become a child

is equivalent to testing children, which the WHO advisers have rejected (Section 9.1). However, if parents are undecided but would consider abortion, it may be best to respect their autonomy and to perform the test, recognizing that they may change their minds after receiving the results.

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) perceived seriousness of the genetic condition within the culture, 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 test results indicating the presence of a potential disability, 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 selective 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).

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: ihere is no universally agreed upon definition of "serious". In a survey of almost 1500 medical geneticists in Europe and the Americas who were asked to list conditions that they considered serious and conditions that they considered not serious (Wertz et al. 1995), the majority of almost 600 conditions listed, including Huntington disease, Tay-Sachs, Down syndrome, cystic fibrosis, and cleft lip/palate, turned up on both lists. What one professional considered serious, another regarded as not serious. Some respondents apparently thought that late onset after a healthy life (Huntington disease) might render a condition not serious, while others may have thought that a child's early death (Tay-Sachs) removed the burden of care from the parents and therefore made the disease less serious than one requiring lifelong care. Cleft lip may not be serious if affordable treatment is available, but could be serious in a nation where most people cannot afford treatment.

Nor is there any definition of what may be considered serious in the future. Conditions and their consequences that were once frequently fatal in early childhood (such as cystic fibrosis) are now medically treatable (but still not curable) and more socially acceptable, and many individuals in developed nations 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. However, 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 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). 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). However, 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, professionals 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 treatable although perhaps not curable. 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 paediatric 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 professionals 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 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 tests 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 yet be diagnosed before birth. Identification of fetal sex and selective abortion of male fetuses that are at 50% risk may enable the parents to avoid 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. With increasing access to gene specific tests, this use is becoming less important.

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, would not ordinarily 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). There

appears to be a trend in most countries towards a willingness to consider requests for sex selection or offer referrals (Wertz and Fletcher, 1998). 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, many 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. Other male relatives can perform these religious duties.

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 disorders in the fetus and undermines the major moral reason that justifies prenatal diagnosis and selective abortion - the prevention of genetics disorders. 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).

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, Wertz and Fletcher, 1998), and to establish a moral climate against sex selection. Sex selection for cultural reasons is not a medical service; professionals do not have to accede to requests or offer referrals. In cases where a professional 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 professional 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. In some cases, there may be medical reasons for the procedure, including the mother's mental health and the fetus's genetic status.

Prenatal paternity testing does not pose the same degree of adverse consequences for society as sex selection. 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 were 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). Chromosomal and single gene disorders account for only a minority of disabilities present at birth.

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.

13. Prenatal Diagnosis: Optimal Provision of Services

Prenatal diagnosis should be provided in a supportive, non-coercive atmosphere that allows couples to make the choices that are best for them in view of their values and parenting goals (Council of Europe, 1990). This includes pre- and post-test counselling, full disclosure of test results, and availability of legal, affordable abortion services.

Counselling is particularly important prior to prenatal diagnosis to secure fully informed choices. Pre-test counselling makes post-test counselling (for those with an affected fetus) less difficult

because prospective parents are better prepared. Proposed guidelines for counselling prior to prenatal diagnosis are given in Table 9.

13.1 Pre-Test Counselling

13.1.1 Content of Pre-Test Counselling

The principles of respect for persons and non-maleficence require that women know the purpose of the tests that they are being offered. This applies to all forms of prenatal diagnosis. A woman and her family should know, before a blood test for maternal serum alpha-fetoprotein, that this test may be the first step on the road to a decision about abortion. She should have the right and the power to refuse such testing if she does not wish to face such a decision. Ideally, all decisions about testing should be couple decisions. If a couple cannot agree among themselves, the woman should make the final decision, because it is her body that is involved.

Pre-test counselling need not always be elaborate. Too strict demands for counselling could be a misuse of scarce resources. Pre-test counselling should be provided for both high-risk families (advanced maternal age, family history of genetic disorder, previous child with a genetic disorder, suspect clinical or laboratory findings in pregnant women) and low-risk families (routine biochemical screening or MSAFP testing). Ideally, both groups should receive counselling covering the topics above. In practice, it may be necessary to abbreviate the counselling for those at low risk.

Women (and their families) receiving ultrasound should also receive similar counselling before the procedure, but the counselling should also explain that ultrasound may identify conditions that can be corrected or ameliorated before birth.

Pre-test counselling has practical advantages in the provision of genetics services. It makes post-test counselling (for those with test results indicating the presence of an affected fetus) much less difficult because couples are somewhat prepared. It helps to prevent unexpected emotional crises. It raises the level of individual or couple awareness and facilitates communication between professionals and couples or individuals.

Pre-test counselling may not be possible for primary care physicians who have many patients and limited time. Physicians need not do basic counselling themselves. Trained paramedics, written material, and audio-visual materials could be sufficient.

13.1.2 Timing of Pre-Test Counselling Relative to Prenatal Diagnosis

Scheduling prenatal diagnosis immediately after counselling reduces the likelihood that a woman will abstain from the procedure. In order to avoid coercion, it may be preferable to provide the possibility of a waiting period of perhaps one to seven days between counselling and prenatal diagnosis. This can pose a hardship for women who must travel long distances, however. In order to avoid this hardship, it may be best to offer women who have traveled to a clinic for counselling the choice of having the procedure the same day. An alternative would be for a community-based counsellor to travel to the family's home or neighbourhood health centre to provide the pre-test counselling. The woman could then decide whether to travel to the clinic for prenatal diagnosis.

Table 9. Proposed Guidelines for Counselling prior to Prenatal Diagnosis

Counselling should include the following points as a minimum:

1. Name(s) and general characteristics of the major disorder(s) that the test may identify. The list of disorders need not be exhaustive. The characteristics of the disorder(s) should be described also in terms of their effects on the future child, on the parents, and on family life.

2. Possibilities for treatment of the disorder(s) after birth and availability of supportive care.

3. Description of the likelihood (risk) that the fetus may have the disorder(s). Risks should be expressed in several ways (as a percent, as a proportion, and verbally).

4. The possibility of unfavourable test results or of fortuitous or unexpected findings.

5. Alternatives available for those with an affected fetus, for example, carrying the fetus to term and caring for the child at home; placing the child in an institutional setting, if available; placing the child for adoption; termination of pregnancy; prenatal treatment for the fetus or early treatment after birth.

6. The possibility of ambiguous laboratory or ultrasonography results.

7. Information that, because most conditions diagnosed in the fetus cannot be treated before birth, knowing about the existence of a condition may not help the fetus.

8. Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth, or are not tested for unless the family is known to be at high risk.

9. The medical risks to fetus and mother posed by the testing procedure.

10. Non-medical risks, if any (e.g., to parental employment or health care, where applicable).

11. Information that non-invasive screens used early in pregnancy, such as maternal serum alpha-fetoprotein (MSAFP), may be the first step on the road to prenatal diagnosis and a possible decision about abortion.

12. Costs of the test and sources of reimbursement for the mother or couple, if applicable.

13. Names and addresses of genetic support groups or organizations for persons with genetic disorders, that people can contact if they wish.

13.2 Full Disclosure of Test Results

Medical/genetic results: all test results relevant to genetic disorders or fetal malformations should be disclosed. These include sex chromosome abnormalities and disorders that may not be considered serious.

Results not relevant to health: sex in itself (in the absence of an X-linked disorder) is not a disease and need not be disclosed. Cosmetic characteristics (height, weight, etc.), in the absence of a genetic syndrome, should not be revealed if these became prenatally diagnosable. Sexual orientation (see 8., above) need not be revealed if this ever becomes prenatally diagnosable. Disclosure of fetal characteristics that are within the realm of normal may lead some families to

use abortion for purposes of cosmetic selection. This practice should be avoided because it could lead to a redefinition of normalcy.

Ambiguous or conflicting results: ambiguous or conflicting test results should be disclosed. Although uncertainty may cause anxiety, it is better to disclose an ambiguous result before birth than to have the patient face an unexpected surprise after birth. New or controversial interpretations of test results should be disclosed.

Normal test results: all normal test results, including those from maternal serum alpha fetoprotein measurements and two or three marker testing, should be disclosed promptly, because testing arouses anxiety in many people.

Disclosure to husband or partner: although both parents should ultimately know the test results, priority should be given to informing the woman. The fetus resides in her body. She should have control over information about both her body and her fetus. If she has difficulty telling her husband, the physician or counsellor should work with her toward the solution that will provide least harm to all concerned.

In some cases, a woman may ask that her husband be told the results first. This request should be honoured, but the medical geneticist has the responsibility to make sure that the woman is acting voluntarily, and that she receives the information in a timely fashion.

Disclosure to a couple's other children: many parents wonder whether to disclose prenatal test results (or even the fact that they have been tested) to their affected or unaffected children. The benefits and harms of disclosure will vary in individual cases. This is a decision best left to the parents. The professional should not tell a couple's minor children, but should be prepared to discuss with the couple the potential benefits and harms of disclosure to children.

Timing and method of disclosure to parents: in order to maximize a couple's options, speed is of the essence. All disclosure of unfavourable test results should be in person, to allow maximum support and counselling. In practice, this may, on rare occasions, be impossible. The benefits of in-person counselling may be outweighed by the anxiety of waiting. If there is a strong need, basic information can be transmitted sensitively by telephone, followed by a clinic appointment. This information should be conveyed only to the woman, however. In rural areas where people may have to travel long distances to a clinic, more of the counselling may have to be by telephone, taking care to insure privacy. If a telephone is not available, it may, very exceptionally, be necessary to use other means for prompt communication, such as adequately trained rural community health workers who could visit the woman's home to deliver the basic information.

13.3 Post-Test Counselling after Findings of a Genetic Condition

Full information about the disorder: post-test counselling should include a description of the full range of severity of the disorder, from least to most affected, and a description of the most usual symptoms characterizing people with the disorder. These symptoms should be described in terms of their functional effects rather than in medical terms. Counselling should describe how a person with the disorder develops over the entire life course, from birth to death. If affected persons themselves experience physical pain or suffering, the counsellor should make this fact clear. The counsellor should describe the possible range of effects of the disorder on family life (including the marriage), as well as financial and emotional costs, possibilities for treatment, education, and supportive living in special settings or in the community. If the counsellor offers

referral to families who have children or siblings with the disorder, care must be taken to offer a sufficient number to represent different parental views and different degrees of severity of the disorder, if relevant. The counsellor may also present the option of carrying the child to term and placing it for adoption as an option, if adoption is a realistic possibility.

Counselling both parents: a couple should be offered counselling together. However, the mother may be seen alone if she desires. At the outset of counselling, the counsellor should explain to both parents that they should not feel guilty. Their actions did not cause the disorder, nor did it result from the woman's or the man's behaviour before or during pregnancy. It is especially important that this information reach the husband, in order to prevent blame falling upon the wife. Counselling should be accompanied by some form of ongoing evaluation that enables the counsellor to see whether the couple actually understands the information provided. There should be evidence of full understanding before the woman or couple is encouraged to make a decision.

Counselling when parental behaviour leads to birth defects: when parental behaviour (e.g., maternal smoking, drug or alcohol abuse, failure to stay on the PKU diet, or physical abuse by a woman's partner) has led to abnormalities in the fetus or child, it may be counterproductive to make the parents feel guilty. Although the fetus or child is damaged, this is not the same as child abuse and should not be referred to legal authorities. Usually the mother had diminished control over her body, especially if she was addicted. The goal of counselling should be to prevent further damage to the fetus or child. This may mean education of the parents, offering the possibility of abortion, offering a supportive environment, on a voluntary basis (preferably a residential institution), where the mother can continue her pregnancy without drugs or alcohol and on the proper diet, or providing support services for the family and the child.

Abortion counselling: for women considering abortion, the counsellor should describe the various methods of abortion available and the attendant risks and discomforts of each. In countries where abortion is legal, methods should be available, and reimbursed by health care systems, on the basis of minimum discomfort and complications for the woman rather than convenience for the doctor.

If a woman chooses abortion, she should be made aware that, while most women in Western nations recover emotionally and return to their usual activities within a month, some feel lingering grief and a few undergo clinically significant depression (Black, 1993; Tunis, 1992). She should be told of the availability of counselling or support groups.

Timing of abortion relative to counselling: a waiting period of at least a day between counselling and abortion is desirable, for several reasons. It allows the woman and partner some time for deliberation after the initial shock of receiving test results. It reduces the possibility of regretting an over-hasty decision.

On the other hand, some women must travel long distances to clinics and cannot afford to spend an extra day near the clinic. In view of these potential hardships, which affect many people, a flexible policy seems best. A mandatory waiting period could impose undue hardship. The counsellor should suggest that a couple take some time to come to a decision. Supports should be available, in the form of inexpensive, subsidized lodging near the clinic, for those who need time to reach a decision. However, prompt abortion services should also be available if a woman needs or wishes them. No woman should have to wait more than a day after she has decided to have an abortion.

14. Termination of Pregnancy Following Prenatal Diagnosis

14.1 Respecting Different Cultural Perspectives

There are many different cultural perspectives about when human life begins. Given the diversity of views, it is unlikely that there will ever be universal agreement on this issue. Therefore, it is best to proceed on the basis of acknowledgement of, and respect for, the views of others. This means that abortion procedures should be available, if legal, even if only acceptable to or used by a minority of a nation's people. Such procedures should be supported by public health funds and provided free of charge. No woman should be coerced into having any procedure; neither should she be coerced into carrying a child to term. Provision of abortion should not become a rationale for reducing support and services for people with genetic conditions.

The following discussion centers on abortions following a pathological finding at prenatal diagnosis. It is difficult to separate completely the issue of termination of pregnancies with an affected fetus from abortion on social grounds or abortion on request, because in most nations there are no medical standards for hereditary disorders or fetal malformations that may warrant abortion. Instituting such standards in pluralistic societies could be oppressive, because different cultural groups may hold different views about the relative seriousness of different conditions. Setting medical standards for "seriousness" of hereditary disorders in the context of prenatal diagnosis and abortion would also place the balance of power in the hands of politicians and administrators, instead of women and couples. The most ethical approach therefore is to leave genetic abortion within the wider context of abortion on request, and to let women and couples decide upon the seriousness of a condition, in view of their personal and social situations.

Nations that have laws forbidding termination of pregnancies with an affected fetus have the obligation to examine the conditions under which prenatal diagnosis is offered. Offering prenatal diagnosis without the possibility of safe, affordable abortion may cause some women to have unsafe illegal abortions. Categorical prohibition is contrary to the premise under which prenatal diagnosis is offered, namely, offering people reproductive choices. A professional who performs prenatal diagnosis in a country where abortion is illegal is ethically obligated not to abandon women with abnormal findings. In nations where abortion is illegal, physicians who perform prenatal diagnosis owe the woman help and support for her choices after receiving results.

In degree of controversy, termination of pregnancies with affected fetuses outranks any other ethical problem in prenatal diagnosis. However, far fewer persons are adversely affected, compared to those harmed by no access to services. Also, the incidence of abortions for genetic reasons is no more than 1% of all abortions (Wertz and Fletcher, 1989a), vastly fewer than elective abortions due to social causes, failed contraception, or personal reasons. Some women choose to carry to term after hearing of genetic abnormalities (e.g., in disorders such as cystic fibrosis) (Wertz et al, 1991). Abortion choices are, however, a special source of emotional suffering, for the reasons shown in Table 10.

Table 10. Why termination of a pregnancy with an affected fetus is difficult

1. The choice usually involves a wanted pregnancy.
2. Many people attribute a higher moral status to the fetus at mid-trimester and at viability.
3. Many parents, who have already viewed the fetus on ultrasound, will have endowed it with the qualities of a living child.
4. There is a wide spectrum of severity in some chromosomal and Mendelian disorders and prenatal diagnosis usually does not predict severity.
5. Improved treatments for some disorders have led to longer life spans for some affected persons.
6. Knowledge that a termination of pregnancy with an affected fetus had occurred could harm the mental health of living children (siblings of the fetus), who have the same genetic condition.

14.2 Difficulties of Abortion Choices

Most pregnancies that proceed as far as prenatal care and prenatal diagnosis are "wanted" pregnancies, even if they were not wanted or intended at the time of conception. There are different degrees of wantedness, but usually by the time a woman receives a second trimester prenatal test result she has started to think of herself as a mother. This may be why many women who would not hesitate to abort an unwanted pregnancy for personal reasons feel emotional pain and guilt about aborting because there is something wrong with the fetus. The mother who receives prenatal diagnostic findings indicating the presence of a genetic condition must make her decision on the basis of the fetus's characteristics. She must also live with her decision. If she aborts, she may feel grief similar to that for loss of a child. If she carries to term, she and her family will be responsible for the child's care.

Many people believe that a second-trimester fetus has greater moral status and therefore deserves greater respect than a first-trimester fetus. Some world religions, including Christianity, Islam, and Judaism, have historically placed greater value on the second trimester fetus. Women's experience of pregnancy makes second-trimester abortions emotionally difficult, because the fetus has affirmed its presence by beginning to move.

A woman having prenatal diagnosis may have seen the fetus on ultrasound and may have begun the process of maternal-infant bonding.

Some of the more common genetic disorders diagnosed prenatally, including Down syndrome and sex chromosome abnormalities such as XXY, vary widely among individuals in terms of effects on daily living. Some children with Down syndrome, given maximum educational opportunities and support, may be able to hold unskilled jobs in protected environments or read at an elementary level. Other children with Down syndrome, given the same level of support, may have I.Q.'s of less than 30 and require lifetime institutional or parental care. In many nations, optimum education and support are not currently available, and children with genetic disorders are unlikely to reach their full potential, especially if a family has few resources of its own. Children with Down syndrome do not ordinarily suffer and are often happy individuals. The "suffering", if any, is that of the parents, brothers and sisters. In the United States, studies suggest that families of children with mental retardation have lives as full and as satisfying as families of

“normal” children. This may not be the case in countries without adequate social support. Women making abortion decisions, if fully informed, have to weigh the possibility that the aborted fetus might have had a happy life after birth, against the possibility that the child would have low potential and would require care that the parents are unable to provide.

Improved treatment for some disorders and improved medical care in general have compounded the problem of abortion choices. Not too long ago, the life expectancy of a child with Down syndrome was markedly lower than average; few reached middle age. Now, at least in developed nations, many people with Down syndrome can expect to reach middle age or beyond. This increase in life expectancy has important implications for care. It is not uncommon for parents in their eighties to have total responsibility for the care of children with Down syndrome in their fifties (an age where most persons with Down syndrome will have developed Alzheimer disease). When the parents die, the care usually falls on the siblings of the affected individual. Women making abortion decisions now have to consider that if they carry the fetus to term, they and their partners may be required to care for the child for the rest of their natural lives rather than for a short term.

Couples who are already the parents of a child with a genetic disorder not causing mental retardation, for example, cystic fibrosis, are frequently concerned that by aborting a fetus with the same disorder they are rejecting their already living child. They may be concerned that if the child were to know about the abortion, the child will have lower self esteem or feel worthless. Careful counselling about if and how to inform the child can overcome this potential problem.

Some groups, especially some of those representing persons with disabilities, have expressed concern that abortion of genetically affected fetuses will direct societal attention and resources away from caring for living persons with genetic conditions, or will obscure environmental causes of birth defects. These concerns have already been addressed under 12.5.

In view of the psychological distress that abortion choices present for women, follow-up is in order for all women who receive prenatal diagnostic results showing the presence of a genetic condition, whatever their decision. Bereavement therapy or support groups should be available, if women request it.

14.3 Twin and Other Multifetal Pregnancies

Ethical problems arise after prenatal diagnosis of one abnormal twin or in multifetal pregnancies where the number of fetuses threatens the mother's ability to carry them all to a point of survival. In the former cases, parents may desperately want to have a normal child but are unable to care for a child with a disability. The latter cases, also marked by desperation, usually follow infertility treatments, including IVF (Evans et al, 1988). Families using IVF for genetic reasons should be told, before initiating an IVF programme, that the procedure may result in a multifetal pregnancy that may require a decision about fetal reduction. Both situations call for a position to do the least harm in a "lifeboat" type of ethical emergency. The principle of proportionality is clearly relevant here. Selective termination of one twin with a disorder or malformation is ethically more complex than genetic abortion of a single fetus (Fletcher and Wertz, 1993). Risks include dangers to the well being of the presumed normal twin and the mother (i.e., the risk of clotting, haemorrhage, and shock). The means are the same in each case. That is, justified feticide. The act of termination is not morally different, in kind, from abortion because of a genetic condition in a single fetus, although the considerations are more complex and the practical procedure more difficult.

14.4 Third-Trimester Abortions

Anomalies are now more frequently discovered in the third trimester because of ultrasound examinations. Decisions about third trimester abortion pose particular ethical difficulties, because the fetus is often viable, albeit with extraordinary medical intervention and reduced likelihood of normal life. There are no cross-culturally acceptable lines of demarcation indicating the severity of the fetal defects for which third-trimester abortion could be ethically allowable. Sometimes the result of denying abortion is a "born fetus" that spends agonized days or weeks in a neonatal intensive care unit before dying (Fletcher et al, 1992). Some have argued that third trimester abortion should be performed only if the fetus has a condition that will be lethal soon after birth and for which no beneficial treatment is available (Chervenak and McCullough, 1990). This argument presupposes that a third trimester fetus is equal to a newborn. This view, however, may be overly restrictive to those who would not place severe obstacles in the way of decision making about third trimester abortions for genetic reasons.

If third trimester abortions are legal, they should be limited to situations for which second trimester abortion was not possible because the fetal condition was not diagnosable in the second trimester. Decisions that can be made in the second trimester are best not postponed until the third trimester.

If abortion is legal in the third trimester, it should be performed in a manner that provides adequate analgesia to the woman, that does not cause the fetus to undergo prolonged suffering, and that does not provoke the woman to change her mind (futilely) during the several days that may be required for dilating the cervix and for vaginal delivery (Hearn, 1990). Procedures that deliver a living fetus that subsequently takes hours, days, or weeks to die are ethically unacceptable; they do not save meaningful life and only lengthen suffering for both fetus and family.

In most cases, fetal therapy will not be a feasible alternative. In cases where therapy is available, however, but involves an invasion of the mother's body, the situation is analogous to Cesarean section, though with potentially greater risks. The mother should have final decision over whether or not fetal therapy is performed. Her body should not be invaded against her will.

15. Preimplantation Diagnosis

Preimplantation diagnosis (PID) offers an alternative to families and societies that wish to avoid abortion. Some users are women who have already had abortions following prenatal diagnosis and do not want to undergo these procedures again. This alternative, however, is costly and may not lead to a live birth. The ethical issues and counselling are similar to those in prenatal diagnosis, except that there is no pregnancy until the fertilized egg is successfully implanted. As there is no worldwide agreement as to when human life begins or when it acquires moral significance, there is no agreement about the moral status of an embryo. Nor is there agreement as to whether discarding an embryo with a genetic disorder, prior to implantation, is the equivalent of abortion. Because some families and cultures regard preimplantation diagnosis as morally preferable to prenatal diagnosis, the option should be offered if a nation has sufficient resources.

16. Keeping Genetically Impaired Newborns Alive

Although most genetic disorders cannot be treated or cured in a primary sense, modern technology has increased the chances of survival for newborns with mental retardation and other

hereditary conditions. As a result, some individuals with severe mental retardation who would formerly have died soon after birth now have significantly longer lifespans. Others die in the first months of life after spending their lives in neonatal intensive care units. Technology, rather than nature, today tends to determine the lifespan. Keeping some infants alive requires extensive medical resources.

When such resources are limited, as they are in many nations, long term intensive care for seriously impaired newborns whose impairments cannot be corrected may have lower priority than treatment of children whose impairments may be overcome with supportive care, education and treatment. Care for seriously impaired newborns may also have lower priority than basic maternal care or well-child care. There is an important ethical distinction between termination of pregnancies with affected fetuses and withholding or withdrawing life supports from a newborn whose impairments are overwhelming, such as trisomy 13 or 18, according to a mutual agreement between parents and professionals. In the former, one is willing to take direct means to end the life of the fetus to prevent its birth. In the latter case, having participated in the decision making around supporting the severely impaired newborn until the prognosis is clear, it is ethically acceptable to forgo life-sustaining measures. Physicians should not withdraw life-sustaining treatment from impaired newborns without the knowledge and agreement of parents. In situations where parents and professionals cannot come to an agreement, an interdisciplinary ethics committee with at least a few lay members may assist the decision makers with a process to explore the options and their ethical disagreements. However, ethics committees must not impose outcomes on decision makers with moral standing to make decisions, i.e., the parents and professionals, and will necessarily work within a country's laws. Individual cases could be discussed in ethical consultations.

A society that keeps a severely handicapped newborn alive by heroic efforts at birth should be willing and prepared to provide the best available support to that child for life. If a society is not willing to support the child, that society should not impose use of heroic methods or lifesaving operations upon doctors or parents unless parents wish it.

17. Protection of Pre-Embryos, Embryos and Fetuses from Environmental and Social Harm

Societies have an ethical responsibility to protect the germ cells, fetus, newborn, and infant from environmental harm. This means a safe working environment for both men and women of reproductive age, equal access to prenatal care, maternal and infant nutrition, and protection from environmental harms, both before and after birth.

17.1 Prenatal Care

The developing embryo is most vulnerable to environmental assaults in its first three to four weeks, during organogenesis. Often substantial damage occurs before the woman even knows that she is pregnant. The need for preventive measures is greatest at this time. The use of simple, cost-effective care before or early in pregnancy could eliminate needs for costly technologies after the child is born. As described above (Part 1, Sections 4.7 and 4.8), prenatal care can uncover social as well as medical causes of ill health. These may include homelessness, alcoholism or drug abuse. The social cannot be separated from the medical aspects of care. Prenatal care should be available to all women, regardless of geographical location or ability to pay.

17.2 Maternal-Fetal Conflicts

Sometimes a pregnant woman's behaviour endangers the life of her fetus (e.g., if a woman with maternal PKU does not go on a low phenylalanine diet). In most cases, an adversarial approach (use of the courts or forcible institutionalization) is unlikely to lead to the best outcome for the fetus. Usually the interests of the mother and the fetus coincide. Most women who intend to carry a pregnancy to term want whatever they think is best for the baby. Apparent conflicts between maternal and fetal interests arise either from (a) poor communication between professional and patient, or from (b) a mother's having lost control over her own body as a result of alcohol or drug addiction. Forcible hospitalization is a last resort that should be avoided because it sets a dangerous precedent for societal control over pregnant women.

17.3 Maternal Employment

Regulations requiring that a worker be shifted to a less hazardous job as soon as she learns that she is pregnant are not sufficient to prevent fetal damage. The fetus is at greatest risk before its existence becomes known.

The most ethical solutions to the dilemma between women's needs and rights to work and the fetus's entitlement to protection are social solutions: sex education, availability of contraceptives, occupational safety and health measures, and liberal paid maternity leaves. If every pregnancy were planned, and if women had a right to return to their former jobs after paid maternity leaves, there would be much less conflict between maternal and fetal interests. Women could tell their employers that they intended to become pregnant, could receive maternity leave (before pregnancy, if the job is hazardous to the fetus) and could return to their jobs without penalties.

18. Research Issues

18.1 Informed Consent

As described above under Informed Consent, all participation in research should be voluntary and should follow established procedures for informed consent. Participation or refusal of participation in research should not affect a person's health care in any way. If research involves children or fetuses, the parent or guardian should give consent (see 9. Testing Children and Adolescents, above) with the knowledge and assent of the child if the child is able to understand.

Individuals participating in genetic research projects may be required to provide a family history. Providing a family history in a research context is different from giving a family history to one's physician in the context of clinical care, because the researcher is not acting on the individual's behalf for diagnosis, prevention, or treatment. In research, the question arises of whether the relatives whose presumed history is provided by another party should themselves be required to give informed consent. This is an unsettled issue in process of exploration (American Society of Human Genetics Executive Committee. Membership Alert, March 28, 2000) On the one hand, in some cases information may stigmatize the relatives, especially if it involves mental or behavioural disorders. Even if names are removed, it may still be possible to identify relatives of a research participant by tracing relationships. On the other hand, the logistical problems of identifying and finding relatives so that they can give consent may be insurmountable. Ethical review committees must take these issues seriously, and, for each research project, must weigh the risk of exposure and stigmatization against the practicalities of locating relatives. The generic issue is security of data in the research project as a whole. Ethics review committees also need to consider the scientific issues involved in use of family histories in research. Data provided by

individuals about their family members may be inaccurate. Locating and requiring family members to provide the information themselves after informed consent could increase the accuracy of information about those who are located. However, many family members may not be locatable, and if consent is required in order to include their information, the comprehensiveness of the dataset may be compromised

18.2 Commercial Involvement and Conflicts of Interest

Traditions of academic and scientific freedom are designed to protect researchers in academic centres, although these freedoms can be threatened by social and political interests. When commercial entities are involved in research, it is particularly important to protect researchers and subjects from possible coercion or pressure to conceal information and findings. Academic institutions that create alliances with industries to conduct research require a strong review process to probe possible conflicts of interest between researchers' scientific responsibilities and business interests (e.g., ownership or part ownership of a company developing a new product). In cases where the review board determines that a conflict of interest may damage the scientific integrity of a project or cause harm to research participants, the board should advise accordingly. Institutions need self-regulatory processes to monitor, prevent, and resolve such conflicts of interest. Prospective participants in research should also be informed of the sponsorship of research, so that they can be aware of the potential for conflicts of interest.

18.3 New and Controversial Research

The clarity of the science of genetics and its tools have led to discoveries that present unique opportunities, e.g., to study the natural history of a genetic disorder. In human genetic disorders, the more knowledge of natural history and the specific genetic mechanisms that cause them, the greater the likelihood of developing diagnosis and therapy. Therapy will evolve both in terms of new drugs to ameliorate the expression of harmful genes and in terms of human gene therapy.

Some disorders literally begin in the embryonic state or very early after implantation. Categorical rejection of research, simply because it may occur in the fetus or embryo, is a reaction primarily from fear rather than rational assessment. Rational approaches to fetal and embryo research are possible, even in societies where sharp restrictions fit better with conservative moral traditions (FIGO, 1993). Every society ought to support national research ethics commissions to debate and recommend guidelines to control possible abuses in fetal and embryo research, as well as to outline standards under which ethically acceptable research can be done. It does not follow that because genetics was abused in the past it is inevitable that genetic information will be abused in the present or future. Societies can build in protections against abuses. After general guidelines for research have been adopted, each proposal can be judged on its own scientific and clinical merits within the national policy.

Closing off an avenue of research prematurely offers little benefit and promotes both social inequality and scientific hypocrisy. Those who can pay (e.g., for fetal tissue transplants) will seek therapy elsewhere. Also, scientists in a nation that suppresses the possibility of embryo or fetal research will use the information generated by others, even when they consider that information to have been derived by unethical research practices.

18.4 Research Involving the Human Embryo

Ought human embryos be utilized for the purpose of research? This question is important from the perspective of scientific knowledge, and it is controversial from the perspective of many of

the world's religious and ethical traditions. Answering the question involves a two part judgement: (1) a moral judgment as to the status of human embryos prior to implantation and (2) a social judgment about the degree of protection in research that should be accorded to human embryos as a class. In making the second judgment and in policy decisions about the question, a crucial factor is how much weight is to be given to potential benefits of embryo research for the health of women, men, and children (United States, 2000).

The embryo does not have the same moral status as infants or children, although it deserves respect and serious moral consideration as a developing form of human life. This judgment is based on three characteristics of pre-implantation embryos: absence of developmental individuation, no possibility of sentience, and a high rate of natural mortality at this stage (National Institutes of Health, 1994).

It is not inconsistent to view the embryo with respect, due to its human origins, and hold at the same time that an experiment ending in an embryo's death cannot "harm" an embryo. The embryo is an organism with human origins, but it is without sentience (feeling) and without interests. Harm cannot be done to such an organism until the capacity for sentience has been established. From this perspective there is a clear difference between the moral status of living children and embryos (Fletcher and Ryan, 1987). To be sure, no society permits comparable experiments with living children who are sentient and who have interests. However, many societies permit investigative or "non-therapeutic" research that does not benefit children in the study as well as taking risks of morbidity and mortality in trials in children with cancer (Furman et al, 1989). It is possible to damage an embryo in research. The damage would become "harmful" in the moral sense only if the embryo was transferred to a human uterus and a future sentient person was harmed by the damage once done to the embryo (Kuhse and Singer, 1990). This possibility can be avoided by regulations forbidding the transfer to a human uterus or any laboratory equivalent of any embryo that has been involved in research.

In terms of the issue of the degree of protection owed by societies to human embryos in research, there is a moderate moral position lying between the polarities of permitting no research and providing no protection. Protection is owed to the human embryo because of its origins and the value of respect for human life. Respect for the human embryo can be shown by (1) carrying out the proposed research first on non-human embryos, (2) accepting limits on what can be done in human embryo research, (3) committing to an interdisciplinary process of prior group review of planned research, and (4) carrying out an informed consent process for human gamete and embryo donors. Although this way of showing respect differs from the position of forbidding human embryo research based on potential for personhood or the genetic integrity of a human embryo, it is closer to that position than a position that human embryos have no moral status at all, or that society has no obligations to regulate human embryo research. Further, respect for the human embryo's limited moral status can be shown by careful regulation of the conditions of research, safeguards against commercial exploitation of human embryo research, and limiting the time within which research can be done to 14 days. This last restriction is in keeping with policy in several nations that permit research with human embryos (Australia, 1984; Great Britain, 1984; American College of Obstetrics and Gynecology, 1986; Human Fertilization and Embryology Authority, 1993; Royal Commission on New Reproductive Technologies, 1993) until the developmental stage when the "primitive streak" appears. At this time, the development of the nervous system begins and the human embryo begins to become a distinct individual.

Those favouring human embryo research are primarily motivated by the prospects of benefits of knowledge about diagnosis and treatment of children, women, and men. Studies of "normal" human embryos will lead to understanding the entire process of fertilization, which cannot be

entirely accomplished by animal research. Additionally, studies of "abnormal" human embryos are a potential source of scientific information at the molecular level about the origins and development of pediatric cancers, malformations and other genetic disorders. Significant weight should be given to the value of this information, especially if it cannot be obtained from animal research or in any other way, e.g., in research on human sperm and eggs. A justification for human embryo research stems from its relevance to pediatric oncology and gene therapy for genetic disorders. To understand the natural history of a disease is a *sine qua non* for optimal approaches to diagnosis and treatment. In these cases, it will be necessary to obtain sperm and eggs from parents who are at higher risk to transmit these conditions to offspring, and to study the genetic mechanisms involved compared to those in "normal" human embryos. Thus, restricting human embryo research only to spare human embryos donated after infertility treatment will not be sufficient. Any argument against human embryo research is obliged to address the social costs to living and future children, and to societies, of foregoing such activities.

Finally, it must be pointed out how illogical and morally self-defeating it is for societies to forbid all research with human embryos and then to require excess embryos resulting from IVF to be discarded at the end of a time period after freezing. Scientists and physicians in these societies will nevertheless use knowledge gained in other societies by human embryo research.

18.5 Fetal Tissue Transplant Research

Many sufferers from neurological disorders, such as Parkinson's disease, may stand to benefit from transplants of fetal cells. Tissue from fetuses spontaneously aborted is not optimal for transplants, because it may be macerated, infected, or otherwise inadequate for therapy. Opponents of use of fetal tissue have argued that it will increase the number of social abortions. In reality, no woman has a social abortion primarily in order to donate tissue for research. Use of fetal tissue should be allowed, provided that (a) the woman consents; (b) the woman is not paid for the tissue; (c) the tissue will go to an anonymous recipient, not known to the woman who donates it; (d) the woman has decided upon the abortion before being asked to donate tissue; (e) the researcher is not the doctor who performed the abortion; (f) no third party is paid for the tissue; and, (g) the abortion is not delayed to recover more or better prepared material. Anonymity of the recipient is important, in order to prevent the possibility that a woman might conceive (or be coerced to conceive) a fetus for the purpose of donating tissue to a family member. Fetal tissue may become beneficial in treatment of such widely varying conditions as Alzheimer disease, spinal column injuries, diabetes, and Hurler syndrome.

18.6 Researchers' Relations with the Media

Researchers have a responsibility to make sure that the public is accurately informed about results without raising false hopes or expectations. Researchers should take care to avoid talking with journalists or reporters about preliminary findings. Sometimes the media report potentially promising research that subsequently cannot be validated. Sometimes the media report research on animals in such a way that the public thinks that the step to treatment for humans is an easy one. Retractions almost never appear in the popular press or on television. Therefore it is important to avoid premature reports. The best safeguard against inaccurate reporting is for the researcher to require, as a condition for talking with the media, that the reporter supply a full written or oral version of what will be reported, so that the researcher can make any necessary corrections.

19. Uses of Banked DNA

19.1 Access to Banked DNA

Stored DNA in tissue or blood samples may provide useful information for examination of genetic disorders in families or for research. Information from DNA specimens may be of importance for relatives and not only for the person from whom DNA originates. Therefore, access to stored DNA for family members is a possibility that needs to be considered. While spouses may not have a right of access, their concerns should be considered. Proposed guidelines for access to banked DNA are given in Table 11.

Access to stored DNA, whether in a clinical setting, or a DNA bank, may present a conflict of interests between the individual and marital partner, family, or society (McEwan and Reilly, 1995). In the following discussion, the term "DNA" is used to apply both to the stored cells themselves or the stored DNA, and to the stored information obtained from DNA examinations, even if the material itself has been destroyed. An individual's DNA may be used to predict the later development of genetic disorders, to estimate possible increased risks of common multifactorial diseases, to establish or disestablish biological relationships, to help in a genetic diagnosis or risk estimation for blood relatives, to help in reproductive planning for the individual, the couple, and sometimes relatives at risk (and in rare cases to help the proper authorities to know whether a person's genetic status poses a danger to public safety). Life insurers, health insurers (in those nations where health insurance is a private industry), and employers (especially in nations where the employer pays for health care or health insurance) could use information from DNA selectively to deny insurance or employment, or vice versa, to select the healthiest clients or employees.

DNA is both unique to an individual and shared by other individuals who are biologically related. Therefore DNA should not be considered the "private property" of one individual, though characteristics or health indicators unique to that individual should be kept confidential. It should be possible to inform others who share part of an individual's DNA, namely biological relatives, about their own health risks and also to allow them access to the DNA which is shared property. Preferably a depositor's agreement to this effect should take place before DNA is banked, or national regulations could specify that biological relatives may have access.

If a couple wishes to have children, both parties have an interest in the child's health and, therefore, both parties have a moral right to access to each other's DNA, but this should not be a legal right.

DNA stored in forensic data banks should be accessible to law enforcement agencies, but otherwise should be under the same guidelines as DNA in other types of storage (please see Table 11).

Institutional third parties are unlikely to use DNA to benefit an individual or family. Therefore they should be forbidden from access under any conditions, with the possible exception of law enforcement agencies (provided that other information links a person to a crime) or, rarely, employees in jobs involving public safety.

Table 11. Proposed Ethical Guidelines for Access to Banked DNA

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

19.2 Use of Stored DNA Samples in Research

Existing stored specimens or samples such as those in university or hospital departments or collections of blood spots need not be the subject of new rules for consent or recontact that may be established in the future.

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

- Protection of individuals from possible discrimination by employers and insurers, etc.
- Possible benefits to the individuals from research findings.
- The possibility of multiple uses of the same sample in different and unforeseen research projects.
- Possible sharing of samples among collaborators, including international collaborators and commercial entities.

- Advantages and disadvantages for individuals and researchers of removing all identifiers (including coded numbers) from a sample.
- The possibility of stigmatizing a community even if samples have no individual identifiers.

A blanket informed consent that would allow use of a sample for genetic research in general, including future, as yet unspecified projects, appears to be the most efficient and economical approach, avoiding costly recontact before each new research project. The consent may specify whether or not an individual would permit access to a sample for blood relatives or spouse. All samples should be used with appropriate regard for confidentiality.

Differentiation between "identifiable" and "unidentifiable" samples is complex and is the subject of comprehensive national ethics commission documents (National Bioethics Advisory Commission, 1999; Wertz, 1999)

20. Patenting

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

21. Gene Therapy

Development of new therapies should be a major goal of genetics services. In the future, many therapies will involve manipulation of genetic material. Gene therapy has two distinct forms: (1) somatic cell therapy; (2) germ-line therapy, including therapy on fertilized eggs (Medical Research Council of Canada, 1990; Bankowski and Capron, 1991).

21.1 Somatic Cell Therapy

Somatic cell therapy applies to cells that maintain normal body functions. It does not include egg or sperm cells, their precursors, or fertilized eggs, and does not affect the next generation. A person treated for a genetic disorder with somatic cell therapy can still transmit the disorder to his/her children. There is worldwide agreement that somatic cell therapy is potentially beneficial for treatment of genetic disorders. Such therapy is ethically similar to other therapies used in treatment of disease. Like other new therapies, somatic cell therapy should be employed only after clinical research trials and with fully informed consent of the persons being treated. Somatic cell therapy should be used only for treatment of diseases or disorders. Any proposals to enhance or "improve" normal human characteristics, including intelligence, should be rejected because their consequences are unknown at present. Enhancement presents potentially grave ethical dangers, including misallocation of resources, increases in social inequality, and redefinition of normalcy (Parens, 1998)

21.2 Germ-Line Gene Therapy

Germ-line gene therapy could affect the egg and sperm, their precursors, and fertilized eggs. Someone successfully treated for a genetic disorder with germ-line therapy would not be able to transmit the disorder to her or his children. The potential benefits of germ-line therapy are that (1) treated individuals would be able to reproduce without worrying that their offspring will have the disorder in question, and (2) in future generations fewer children will be born with the disorders to which therapy has been applied (though there will always be new mutations). The potential risks of germ-line therapy are that (1) it could in theory affect the entire constitution of children developing from the treated sperm or egg, in unexpected, harmful, and dangerous ways about which we can only speculate at present; (2) the damage would be irreversible; and, (3) the damage would extend to future generations.

Germ-line therapy is not necessary for the treatment of disease in living persons, but might eventually (over a long period of time), reduce the number of people who would need treatment in future generations. Germ-line therapy could be involved in the treatment of embryos identified through pre-implantation diagnosis (Bonnicksen, 1994).

It may be premature to pass judgment on a therapy without knowing more about its potential risks and benefits. As in other areas of medicine, knowledge will come from research on other species. Future ethical guidelines for use or prohibition of germ-line therapy can only be established on the basis of carefully controlled, long-term research.

21.3 Therapies Involving Expression of Genes

Therapies aimed at modifying the expression of genes appear to have great potential for the treatment of Mendelian as well as multifactorial disorders, and developments in this field should be encouraged. Like somatic cell gene therapy, manipulation of gene expression in the individual is of no consequence for her or his descendants.

Like the preceding categories of genetic therapies, therapeutics modifying gene expression should for the time being only be used for disorders where there is no other efficient and safe treatment.

21.4 Cloning

Research using human stem cells to grow new tissues (in order to repair or replace those damaged by disease) holds potential promise. Some of this research may involve nuclear fusion of an adult individual's cell with an enucleated egg, a first step toward potential human cloning. The possible benefits of research using nuclear fusion to produce tissues for the treatment of disease are recognized, provided that there would be no attempt to reproduce an entire human being. At the present time, " reproductive human cloning" is unsafe and should not be attempted.

22. Summary

There appears to be broad international support for the general ethical guidelines in medical genetics listed in Table 12 among health workers and in societies at large (Bankowski and Capron, 1991; Wertz and Fletcher, 1993; Wertz, 1997). Respect for persons underlies all statements in Table 12. Respect for persons includes informed consent, right to referral, full disclosure, protection of confidentiality, and respect for children and adolescents in the context of genetic testing.

Table 12. Review Of Ethical Issues

1. Existing genetics services in a nation should be available equally to everyone regardless of ability to pay and should be provided first to those whose need is greatest.

2. Genetic counselling should be non-directive.

3. All genetics services, including screening, counselling, and testing, should be voluntary, with the exception of screening newborns for conditions for which early and available treatment would benefit the newborn.

4. All clinically relevant information that may affect the health of an individual or fetus should be disclosed.

5. Confidentiality of genetic information should be maintained. When there is a high risk of serious harm to family members at genetic risk, the information should be used to avert this harm. If the individual refuses to tell her/his family, the professional may consider overriding confidentiality.

6. Individual privacy should be protected from institutional third parties, such as employers, insurers, schools, commercial entities, and government agencies.

7. Prenatal diagnosis should be performed only for reasons relevant to the health of the fetus and only to detect genetic conditions or fetal malformations.

8. Choices relevant to genetics services, including choices about counselling, screening, testing, contraception, assisted procreation where culturally accepted, and abortion following prenatal diagnosis, where legal, should be available on a voluntary basis and should be respected.

9. Optimum support and education should be provided for children and families with genetic conditions.

10. Adopted children and others with biological relationships outside the family should be able to receive information about their biological relatives, under strict anonymity rules.

11. Research protocols should follow established procedures for review and informed consent.

12. Protocols for experimental human gene therapy should receive national review, with attention to the potential benefits or risks arising from various approaches to therapy.

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