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. Guidelines for pre-test counselling appear in Table 7.

**Table 7**

**GUIDELINES FOR PRE-TEST COUNSELLING**

Counselling should include the following 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 individual, on the parents, and on family life.

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

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

4. Alternatives available for those with unfavourable test results (carrying the fetus to term and placing the child for adoption; caring for the child at home; placing the child in an institutional setting, if available; termination of pregnancy).

5. The possibility of unfavourable test results.

6. The possibility of ambiguous results for amniocentesis, chorionic villus biopsy, and ultrasound.

7. Information that the test will not help the baby.

8. Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth.

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. Non-medical information (e.g., fetal sex) that the test may reveal

12. Costs of the test and sources of reimbursement for the patient, if applicable.

13. Names and addresses of genetic support groups or organizations for persons with genetic disorders.
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 pathological test results) much less difficult because counsellors are somewhat prepared. It helps to prevent unexpected emotional crises. It raises the level of counsellor awareness and facilitates doctor-patient communication.

Pre-test counselling may not be possible for primary care physicians who have many patients and limited time. Basic counselling need not be done by physicians 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 is best to offer women who have travelled 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.

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 detectable. 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 physician 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 counsellors 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 Pathological Findings

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

Ideally, a couple should be seen 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. Methods should be offered 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 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. Genetic abortion 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 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.

The following discussion centres on genetic abortions following a pathological finding at prenatal diagnosis. It is difficult to completely separate the issue of genetic abortion for fetal conditions 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 genetically indicated abortions have the obligation to examine the conditions under which prenatal diagnosis is offered. It is especially cruel to offer prenatal diagnosis without the possibility of safe, affordable abortion. 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 county where abortion is illegal is ethically obligated not to abandon women with abnormal findings. The professional owes such women a referral to services providing safe, affordable abortions, outside the country if necessary.

In degree of controversy, genetic abortion 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 genetic 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 do not choose abortion 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 8.

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<td>WHY ARE GENETIC ABORTION CHOICES DIFFICULT?</td>
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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.

5. Improved treatments for some disorders have led to longer life spans for some affected persons.

6. Knowledge of genetic abortion 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 unfavourable prenatal diagnostic findings 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. 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 or unwilling 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 many persons with Down syndrome will have developed Alzheimer's 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 unfavourable prenatal diagnostic results,
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). Risk of dangers to the well-being of the presumed normal twin and the mother (i.e., the risk of clotting, hemorrhage, 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 genetic abortion, although the considerations are more complex and the practical procedure more dramatic.

14.4 Third Trimester Abortions

Anomalies are now more frequently discovered in the third trimester because of high resolution 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 must not be 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 (futily) during the four to five 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 and less likelihood of successful outcome. 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 offers an alternative to families who oppose abortion. 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 regard preimplantation diagnosis as morally preferable to prenatal diagnosis, the option should be offered if a nation has sufficient resources. However, because the option is costly and has a lower success rate than other options (e.g., gamete donation, adoption of a child without a genetic disorder), it should not be a priority in a nation's health planning.

16. Keeping Genetically Impaired Newborns Alive

Although Down syndrome cannot be treated or cured in a primary sense, modern technology has increased the chances of survival for newborns with this and other hereditary conditions. As a result, some individuals with severe mental retardation who would formerly have died soon after birth now live nearly normal 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 such 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 should have lower priority than treatment of children who may lead healthy lives or whose impairments may be overcome. Care for seriously impaired newborns should also have lower priority than basic maternal care or well-child care. There is an important ethical distinction between genetic abortion after pathological findings and withholding or withdrawing life-supports from a newborn whose impairments are overwhelming (according to a mutual agreement between parents and physicians). 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 consented to delivery of the fetus and 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 can 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 legal and moral standing to make decisions, i.e., the parents and physicians.

A society that keeps a severely handicapped newborn alive by heroic efforts at birth should be willing and prepared to support 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), 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 doctor 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.

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 lead to discoveries today 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 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 are more fitting 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 judgment: (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.

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 embryo because of its origins and the value of respect for human life. Respect for the embryo can be shown by (1) accepting limits on what can be done in embryo research, (2) committing to an interdisciplinary process of prior group review of planned research, and (3) carrying out an informed consent process for gamete and embryo donors. Although this way of showing respect differs from the position of forbidding embryo research based on potential for personhood or the genetic integrity of an embryo, it is closer to that position than a position that embryos have no moral status at all, or that society has no obligations to regulate embryo research. Further, respect for the embryo's limited moral status can be shown by careful regulation of the conditions of research, safeguards against commercial exploitation of 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 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 embryo begins to become a distinct individual.
Those favouring embryo research are primarily motivated by the prospects of benefits of knowledge about diagnosis and treatment of children, women, and men. Studies of "normal" embryos will lead to understanding the entire process of fertilization, which cannot be entirely accomplished by animal research. Additionally, studies of "abnormal" 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 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" embryos. Thus, restricting embryo research only to spare embryos donated after infertility treatment will not be sufficient. Any argument against 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 them to be discarded at the end of a time period after freezing. Obviously, scientists and physicians in these societies will use knowledge gained in other societies by 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 abortions. In reality, no woman has an 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. **Recommendations for Banking and Using DNA from Families with Genetic Disorders**

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 should 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.

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.

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 9).

20. **Anonymous Testing for Epidemiological Purposes**

Researchers and government health departments should be permitted to conduct anonymous testing or screening on the general population in order to establish the prevalence of genetic anomalies and deleterious genes. Because the testing or screening is anonymous, it does not require informed consent. PCR (polymerase chain reaction) amplification has made it possible to use a single blood spot for multiple tests. Blood spots collected in screening newborns for treatable disorders could be used to provide epidemiologic information about genetic predispositions to disorders of late onset. Care must be taken to ensure that such testing remains anonymous and that results cannot be traced to individuals or families. Similar programmes to establish prevalence of HIV infection in populations have
succeeded in protecting anonymity. In cases where the information derived from blood spots or stored specimens might be useful to individuals, the code of anonymity may be broken.

Table 9

GUIDELINES FOR ACCESS TO BANKED DNA

- Control of DNA should be familial, not individual. All blood relatives should have access to stored DNA for purposes of learning their own genetic status, but not for purposes of learning the donor's status.

- Family members should have access regardless of whether they contributed financially to the banking of the DNA.

- DNA should be stored as long as it could be of benefit to living or future relatives or fetuses.

- Attempts should be made to inform families, at regular intervals, of new developments in testing and treatments. Donors should inform DNA banks of current addresses for follow-up.

- After all relatives have died or all attempts to contact survivors have failed, DNA may be destroyed, or stored for research purposes.

- 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 tell the spouse any relevant information.

- Except for forensic purposes or instances when the information is directly relevant to public safety, there should be no access for institutions without the donor’s consent. Insurance companies, employers, government agencies, and other institutional third parties that may be able to coerce consent should not be allowed access, even with the patient's consent.

- Qualified researchers should have access, provided that strict confidentiality is observed or that identifying characteristics are removed.

- Family members have a moral obligation to help each other and future generations by participating in research related to familial diseases.

- Potentially valuable tissue specimens in departments of pathology that could be useful to families in the future should be saved and should be available.

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.

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.

At present, germ-line therapy is not technically feasible. It 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 also be used in the treatment of embryos identified through pre-implantation diagnosis (Bonnicksen, 1994).

Fear of germ-line therapy is largely fear of the unknown, overshadowed by the abusive historical record of the eugenics movement. It is 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. 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 potentials 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 serious disorders where there is no other efficient and safe treatment.
22. International Guidelines on Ethical Issues in Medical Genetics and Related Areas

There appears to be broad international support for the general ethical guidelines in medical genetics listed in Table 10 among health workers and in societies at large (Bankowski and Capron, 1991; Wertz and Fletcher, 1993). Respect for persons underlies all statements in Table 10. 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.

The following detailed guidelines are derived from the General Ethical Guidelines in Medical Genetics listed in Table 10 overleaf.

- Access to genetics services. Access to genetics services should not depend upon social class or ability to pay. Whatever services exist in a nation should be available equally to everyone. Genetics services should be provided first to those whose need for them is greatest.

- Non-directive counselling. This is already a formally accepted approach around the world. These guidelines, and this document acknowledge its desirability, its difficulty, and its limits.

- Voluntary approach to genetics services, including genetic counselling, screening for susceptibility to common diseases or to occupationally-related diseases, presymptomatic testing, testing children, and prenatal diagnosis. There should be no coercion, whether overt or covert, attached to genetics services. Persons who choose or refuse genetics services should not be the objects of discrimination or stigmatization. Persons who choose or refuse genetic testing or services should not be penalized in terms of health care, employment, or insurance.

The only exception to the rule of voluntary screening should be newborns, if, and only if, early treatment is available that would benefit the newborn. Governments have obligations to protect their most vulnerable citizens. Newborns cannot protect themselves. Therefore, governments may mandate screening for newborns who would be harmed by the absence of prompt treatment. Governments that mandate newborn screening have the ethical obligation to provide prompt, affordable treatment for the disorders for which they screen. Otherwise the screening is in vain.

- Full disclosure of clinically relevant information to patients. 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 or 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. Test results without direct relevance to health (e.g., nonpaternity, fetal sex in the absence of X-linked disorders) may be withheld if this appears necessary to protect a vulnerable party. Disclosure includes the duty to recontact individuals or families if new developments arise that are relevant to health.
Table 10

GENERAL ETHICAL GUIDELINES IN MEDICAL GENETICS

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 as nondirective as possible.

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 except when there is a high risk of serious harm to family members at genetic risk and the information could be used to avert this harm.

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, and abortion following prenatal diagnosis, should be available on a voluntary basis and should be respected.

9. Adoptive children or children conceived from donor gametes should be treated equally with biological children under the guidelines.

10. Research protocols should follow established procedures for review and informed consent. Research on preimplantation genetic diagnosis should be permitted.

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

- Duties to family members. In genetics, the true patient is a family with a shared genetic heritage. Family members have a moral obligation to share genetic information with each other. If children are intended, individuals should share information with their partners.
Individuals have a duty to inform other family members who may be at high risk. If an individual will not do so, the medical geneticist may issue a general warning to family members, but without revealing information about the affected individual. Preserving patient confidentiality is a well-known duty in medicine. This duty is mitigated if it conflicts with another well-known duty, preventing harm to third parties.

- Protection of privacy from institutional third parties. Medical geneticists should recognize the potential for harm when institutions are allowed access to genetic information about individuals, even with the individual's consent. Therefore such institutions should not have access to such data and should not be permitted to require genetic tests.

- Prenatal diagnosis should be performed only for reasons relevant to the health of the fetus or the mother. Prenatal diagnosis should not be performed solely to select the sex of the child (in the absence of an X-linked disorder). Sex selection, whether for male or female, denigrates the fundamental personhood of those already born, and has the power to harm societies by unbalancing sex ratios. The potential harm to large groups of people outweighs any immediate benefits to individuals or families.

Prenatal diagnosis can be used to prepare for the birth of a child with a disability. Therefore prenatal diagnosis should be available to parents who request it but oppose abortion, provided that they understand and are willing to accept the risks to the fetus.

In some cases, prenatal diagnosis may be performed to protect the health of the mother. These include cases of morbid anxiety, clinically confirmed, or situations where prenatal paternity testing would benefit the mother's mental health (e.g., if rape occurred while a couple was trying to conceive).

Professionals should recognize the human and economic costs involved in prenatal diagnosis and should limit its use to situations where there is a clear benefit.

- Respect for and safeguarding of personal and parental reproductive choices. Reproductive decisions should be the province of those who will be directly responsible for the biological and social aspects of childbearing and childrearing. Usually this means the family, which takes many forms around the world. When a couple is unable to reach an agreement, however, the mother should have the final power of decision.

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. Choice is more than the absence of legal prohibition or coercion. Choice should include the economic and social ability to act upon a decision, including decisions to terminate a pregnancy or to raise a child with a disability. There should be a positive right to affordable genetics services, safe abortions and medically indicated care for children with disabilities.

- Adoption. Adopted children or children born from use of donor gametes, and their social parents, should have the right to know whatever medical or genetic information about the genetic parents that may be relevant to the child's health. Genetic testing of adopted children or children awaiting adoption should fall under the same guidelines as testing of biological children.
• Research in genetics. All research should be preceded by full information and informed consent. Persons who are economically or socially disadvantaged should not be used as research subjects to benefit those who are financially better off. Members of all social groups should serve as research subjects in proportion to their numbers in a nation’s population, as far as possible.

Members of families with genetic diseases have a moral duty to participate in important research that could benefit other people with the same health problem, even if it does not benefit themselves. Research on the human embryo and on the use of fetal tissue has potential in developing treatments for genetic diseases. Surplus embryos resulting from in-vitro fertilization, when used in a research setting, should not receive greater legal or moral protection than children. International agreement about procedures that are hypothetical or in the research stage should wait until there is further scientific clarification of the issues and consequences.

• Human Gene Therapy. Somatic cell therapy changes only the treated individual, not his or her descendants. Somatic cell therapy as well as other genetic therapies that have no consequences for descendants should follow the same protocols as other experimental therapies. Germ-line human gene therapy changes not only the treated individual, but also the persons eggs or sperm. Germ-line therapy is still in the hypothetical stage and should be neither encouraged nor forbidden.

* * * * *
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REFERENCES


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ANNEX I

International Bibliography on Ethics and Genetics

Argentina


All of the above papers refer to the following points:

1. In medicine, diagnostic procedures must be followed by treatment and/or prevention.

2. In medical genetics the professional team must consider:
   - Protection of human life from the beginning (conception).
   - Protection of the physical and psychological health of the family group.
   - Accuracy of methods.
   - Respect for the parents’ decisions.

3. If therapy or diagnosis for a genetic disease is not available in our country, medical geneticists must inform their patients of clinics in other countries where these are possible, and make the necessary contacts to help patients to travel.

Brazil


Examines the ethics of experimentation and fraud in science. Transcribes Hippocratic Oath, Nuremberg Code, the Helsinki Declaration, and regulations of USA Food and Drug Administration.

Integrates concepts of genetics and neurophysiology with cultural ones, trying to explain and understand the ethical behaviour.

Each issue considers a special problem. Those already examined were: (a) AIDS and Bioethics; (b) Terminal patients; and, (c) Abortion.

Canada

Medical Research Council of Canada, Guidelines for Research on Somatic Cell Gene Therapy in Humans (Ottawa: Minister of Supplies and Services, 1990). This document defines the scientific and ethical requirements for somatic cell gene therapy research on humans. Guidelines of the Medical Research Council are binding only for the research which it funds, but they are often used in other research settings.

This study looks at the impact that rapidly changing scientific knowledge and philosophical approaches will have on the definition and legal protection of the human genetic heritage. The concept of genetic heritage and the legal means to protect it must be situated within what is meant by inherent human dignity.

Science Council of Canada, Genetics in Canadian Health Care (Ottawa: Minister of Supplies and Services, 1991).
This report reviews the current and potential role of genetic knowledge and technologies in Canadian health care and examines related policy issues.

Privacy Commissioner of Canada, Genetic Testing and Privacy (Ottawa: Minister of Supplies and Services Canada, 1992).
This report examines how Canadians might benefit from the potential genetic technology without undermining our autonomy and our privacy.

Royal Commission on New Reproductive Technologies, Proceed with Care (Ottawa: Minister of Government Services, 1993).
This report addresses the moral, ethical and social aspects of the new reproductive technologies. The issues of prenatal diagnosis for genetic diseases, sex selection and gene therapy receive particular attention.
Chile

The new science of "Bioethics" proposes that ethical codes must be based on scientific and technological progress, using relative parameters instead of traditional absolute values. Bioethics has begun to condemn traditional ethics as a barrier to scientific progress. Crucial points in this controversy are the legal status of abortion, euthanasia and ectogenesis, which reflect a moral crisis of civilization. The crisis of our culture may be solved only if philosophers and theologians join physicians and biologists to discuss codes of medical ethics and the contribution of Christian thinking to the progress of mankind.

The scientific community is becoming aware that mankind has started to directly manipulate the mechanisms that transmit life and guide our species evolution. The biotechnical revolution of the last decades is dismantling the normal processes of biological balance through the early detection and healing of defects, the manipulation of reproduction and the rupture of species boundaries with interspecies hybridization. All these issues open debates about the ethical limits of scientific research freedom. Genetic ethical codes have been proposed that restrain genic therapy methods, genetic heritage manipulation and the patent right for DNA sequences. The Valencia Statement (1990) on ethics and the human genome tries to establish a minimal consensus among scientists.

Czech Republic

Kučerová M et al. Úvod do klinické genetiky, Avicenum, Praha, pp. 311-312, 1981.
The basic monograph about medical genetics. One chapter deals with ethical and psychological problems of genetic counselling, describing the non-directive approach in very general terms.

The major textbook on clinical ethics for medical students. A chapter entitled "Germinal ethics" is devoted to ethical aspects of abortion and contraception, conception and IVF, and genetic engineering.

The patient's position in the former socialist system is compared with that in the new socio-economic situation.

Emphasizes the right to free choice regarding reproductive practice. Genetic screening and prenatal diagnosis should be available, but decisions not to abort must be respected.

Discusses specific problems of prenatal diagnostics, including refusal of testing, prenatal diagnosis of gonosomal aberrations, and balanced translocations. Stresses the need for close cooperation with gynaecologists, obstetricians, and specialists in infertility.
A review of ethical problems of genetic counselling, prenatal diagnostics and DNA analysis. 
Includes Ethical guidelines of the Czech Society of Medical Genetics.

Denmark

Danish Council of Ethics. Fetal Diagnosis and Ethics. Copenhagen, 1990. Includes recommendations for genetic counselling, and discusses overriding ethical considerations such as suffering, handicap, and normality, access to services, and psychosocial aspects of abortion.

Kemp P. Det uerstattelige. En Teknologi etik. (The irreplaceable: An ethics of technology.) Copenhagen 1991. A monograph on many issues within the area of medical genetics and a trendsetter for genetics and ethics.


Both these anthologies are edited by the Danish Council of Ethics. They list a number of fine articles on genetics, genome mapping, and ethics and provide a good overview of Danish research in this area.


The "sociology of ethics": a sociological approach to how norms of acceptability are created.

Egypt

The introduction to these volumes documents ethical guidelines for the provision of genetics services.

France

This book contains important contributions on predictive medicine, prenatal diagnosis, gene therapy and gamete donation by ethicists, philosophers, psychiatrists, anthropologists, sociologists, obstetricians and geneticists.

Empirical survey directed to the counsellors of the Marseille prenatal diagnosis centre showing the role of insurance coverage to reduce socio-cultural inequities for the maternal age reason.


A report to the Prime Minister, which compares the approaches of bioethics issues in different countries in three areas: status of the body, medically-assisted procreation and organ transplantation.


Evokes the recent problems encountered with ICF, prenatal diagnosis, genetic therapy, graft transplant ...

Sève L. Pour une critique de la raison bioéthique. O. Jacob (ed), 1994, 416 pages.
This book is an introduction to the bioethical reflexion.

This chapter presents the rationale for the French legislation that was initiated in the 1990's and which was finalized and adopted in 1994.

Explores the reasons why 291 women, eligible because of their age, did not utilize fetal karyotyping to detect a possible chromosomal anomaly. Access to amniocentesis depended on the physician's and male partner's attitudes towards prenatal diagnosis and abortion. In 11% of the cases, the woman asked for the test but the physician advised against it. The existence of social welfare coverage is also a strong determinant of women's access to prenatal diagnosis. Institutional policies should ensure greater equality of access while allowing for individual preferences.

The impact of prenatal diagnosis varies between European countries, as the indications and the uptake vary. In 2 countries, termination of pregnancy is not available. In the other 11 countries studied, agreement exists on five indications: advanced maternal age, a previous child with a chromosomal abnormality, parent carrier of a balanced anomaly, mother carrier of an X-linked disorder, and malformation at ultrasound. The prenatal diagnosis uptake for mothers above the maternal age limit varied from 10 to 88% per cent. European harmonization of the indications is not considered feasible at present, because of the rapid changes in prenatal diagnosis policies even within countries.
This is the report on which the French Bioethics law, voted on July 29, 1994 was based. It contains a clear description of the techniques already available, of public policies in France and in other countries, and discusses in depth all the ethical issues raised by prenatal diagnosis, medically assisted procreation and predictive testing. A list of unacceptable objectives includes Eugenics as a public policy, sterilization of mentally handicapped persons, a non-supportive attitude toward handicapped persons and their families, non-resuscitation of disabled persons, and fetal sexing exclusively in order to choose the sex of the baby.

Germany


Stresses the ethical aspects of human genome analysis considered in different contexts, and brings together different European perspectives. The volume is a collection of papers from the workshop "Ethics of Human Genome Analysis -- Survey of the European Discussion", which took place at the University of Tübingen, Germany, from 11-14 September 1992.

Hungary

Suggests possible revisions of law to discourage births of children who will not be healthy. Includes many interesting case studies.

Italy


Comitato nazionale per la Bioetica. [Italian Committee of Bioethics]. Documento sulla Sicurezza Delle Biotecnologie [Statement on Biotechnology]. Presidenza del Consiglio dei Ministri Dipartimento per L’Informazione e L’Editoria.

Comitato nazionale per la Bioetica. [Italian Committee of Bioethics]. Problemi della Raccolta e Trattamento del liquido Seminale Umano per Finalità Diagnostiche [Problems in use of human semen for diagnostic purposes]. Presidenza del Consiglio dei Ministri Dipartimento per L’Informazione e L’Editoria.

Japan


Mexico

Discusses some ethical aspects of prenatal diagnosis (mainly amniocentesis), screening programmes for genetic diseases, and gene therapy in the light of technological developments.

Norway

Berg K. Ethical problems arising from research progress in medical genetics. In: Research Ethics (eds. K. Berg & K.E. Tranøy). Alan R. Liss, Inc., New York, 1983, pp. 261-275. The paper is a contribution to an international research ethics meeting in the Norwegian Academy of Science, and gives a state-of-the-art review of research in medical genetics as of the time of the meeting. It is argued that although the application of new genetic knowledge in practical medicine poses ethical challenges, most of the problems can be adequately handled within the framework of traditional patient-physician relationships (traditional "clinical ethics"). The need for strict data protection concerning genetic tests and future disease risks is emphasized.

Berg K. Ethical issues in molecular genetics. In: Ethical Issues of Molecular Genetics in Psychiatry (eds. Srám RJ, Bulyzhenkov V, Prilipko L & Christen Y). Springer-Verlag (Fondation IPSEN), Berlin, 1991, pp. 1-11. Potentials and limitations of DNA technology in diagnosis, disease prevention and ultimately in therapy are discussed in this paper. It is pointed out that preimplantation genetic analysis of fertilized human eggs for certain rare conditions would have the advantage of avoiding problems with respect to abortion following prenatal diagnosis. The need for rational decisions concerning future use of fetal tissue for treatment of serious disorders is emphasized. It is stated in the paper that an operational definition of "bioethics" as relating to man should include that it is a system of consistent thinking aimed at resolving conflicts between, on the one hand, needs with respect to research, patient treatment or disease prevention, and, on the other hand, the need or desire to protect people from harm or significant discomfort.


It is stressed in the paper that the unique characteristics of genetic disorders, i.e., that a disease in one person may affect the health prospects of other persons (in the family), could require other laws or regulations than those which are adequate for diseases affecting only a single individual. The "patient" may be an entire family rather than just one single person. In order to counsel one member of a family it may be necessary to have detailed information about several other family members. It is argued that information about future disease risks in persons who today are healthy should be handled differently than data on previous or current diseases. Knowledge about a person's future disease risk must not be used to the person's disadvantage, and genetic information about a person's future disease risk must not be available to employers, insurance companies, credit institutions, educational institutions or other traditional consumers of health information.


The situation in Norway is reviewed with respect to medical genetic services and societal attitudes in Norway, with special emphasis on public debate on themes relating to prenatal diagnosis and induced abortions.
Poland


Code of Medical Ethics (passed by III Congress of Polish Doctors, 12-14 December 1993; Article 37: Procreation: A doctor is obliged to inform patients about the diagnostic and therapeutic options of contemporary medical genetics, including prenatal diagnosis. Article 28: A doctor and his coworkers are obliged to secure confidentiality of the stored genetic material (DNA samples) of patients and members of their families.


The Russian Federation

Baev AA. When the genome is deciphered. (Russian). Chelovek. 1991. No. 3. An overview of ethical implications which will arise when wide-scale use of invasive procedures based on genomic technologies become available.

Ivanov, VI. Human genome research and consensus ethics in medical genetics. In: Ethics and Human Genetics. Council of Europe Press. Strasbourg. 1993, pp. 85-86. Due to the broad ethnic and cultural diversity of humanity, general civil and medical ethical codes should be supplemented with consensus-based items considering newly introduced technologies for family planning and reproduction.


South Africa

This chapter covers topics such as genetic counselling (and its aims), for both genetic disorders and prenatal diagnosis; the benefit of screening at risk populations for specific genetic disorders; genetic engineering and cloning; fertilization and embryo manipulation; the new eugenics and the necessity for control on sex selection.

This chapter covers ethical issues in: taking family histories and informing family members; diagnostic accuracy; risk information, perception, evaluation and counsellor bias; to treat or not to treat genetic disorders, such as Down syndrome, spina bifida and cost-benefit in treating phenylketonuria; prenatal detection of heterozygotes; antenatal diagnosis and selective termination of pregnancy; and individual solutions for individual problems.

Topics include prenatal diagnosis and selective abortion with reference to some South African examples and the South African Abortion and Sterilization Act of 1975 (which legalised abortion for a limited range of indications including genetic disorders); research in medical genetics with reference to the South African Guide to Ethical Considerations in Medical Research; periconceptional vitamin supplementation and neural tube defects; the XYY project; the ethics of distributive justice in improving health and welfare in Southern Africa.

Topics covered in this chapter include preventative and predictive medicine (e.g. the controversial carrier screening for CF, predictive testing for Huntington disease and carrier tests for Tay-Sachs disease); new carrier reproductive strategies; gene therapy in somatic cells and germ line; the new eugenics age which requires constant vigilance; the human genome project; the motives and social responsibilities of scientists, the most fundamental value being respect for the dignity and worth of the individual; and the necessity to take more interest in the third world.

This paper covers the benefits of DNA technology and the difficulty in spreading these benefits equitably, especially in South Africa; inequalities in health care; and the response of the medical profession historically and at present in South Africa.

Sweden

The recommendations of a parliamentary commission studying the medical, legal, ethical problems of applications of genetic research on plants, animals and man.
Göran Hermerén, Gentekniken och människan: Nya etiska utmaningar, Tankar om Gen-Etik i människans, djurens och naturens värld, Stockholm: Hybrid-DNA-Delegationen, 1993, pp 7-44.  
An attempt at a systematic survey and analysis of the major ethical challenges of gene technology to human beings.

Document from a conference on the future of genetic research and its consequences for individuals and society with ideas concerning possible research initiatives.

Att spåra sjukdomsanlag - prediktiv gentestning. Etiska vägmärken 7, with contributions by e.g., Gunnel Elander, Göran Hermerén, Håkan Olsson and Jan Wahlström, Stockholm: Statens medicinsk-etiska råd, 1994.  
Using different examples as point of departure (Huntington disease, breast cancer, cystic fibrosis, etc.), this book contains discussions of medical, empirical and ethical problems raised by presymptomatic diagnostics.

These booklets present in a popular way the most significant advances in today’s biomedical research and discuss the effects they may have on plants, animals and human beings as well as the ethical problems they raise.

**Turkey**

New techniques offer the possibility of treatment or prevention for some genetic diseases, but cause some problems that are difficult to solve with classic ethical rules. In the future, society’s perceptions of the facts will solve some ethical problems.

Reports results of a 1985 survey of the ethical views of Turkish geneticists.

Bökesoy I, Arda B. İnsan Genomu Projesinin (HUGO nun) Etik ve Sosyal Yönleri. (Ethical and Social Aspects of HUGO. Medical Ethics, 1:22-26, 1993.)  
HUGO is one of the biggest projects of the twentieth century. It aims at improving the health of future generations. The authors discuss ethical and social value problems caused by this research.

The evolution of modern genetics and the HUGO project are briefly presented in relation to ethical outcomes and to interventions in nature. The author discusses eugenics, especially debates on gametic selection in Turkey, counselling, privacy, access to and cost of genetic technologies.

The authors discuss the commercial invasion of sex selection firms in Turkey. Statements by the owner of such a firm and his use of procedures without permission prompted new legal and ethical considerations. The authors emphasize that sex is not a disease and does not need a treatment. Outcomes of selection, if permitted, are discussed in regard to family and economics. If selections occur, there will be a need for a committee to control them. The Ministry of Health is organizing a committee.


In Turkey, abortion laws require permission from parents. The author discusses outcomes for the mother who will carry the baby and care for it afterwards.

UK

Report of Working Group of the Royal College of Physicians Committee on Ethical Issues in Medical Genetics.


Nuffield Council on Bioethics -- Genetic Screening -- Ethical Issues -- December 1993 published by Nuffield Council on Bioethics, 28 Bedford Square, London WC1B 3EG. This is the most important recent publication. It is a report from a Working Party on Genetic Screening chaired by Professor Dame June Lloyd and is a comprehensive coverage of the area.


USA


Covers most ethical issues in depth, including limitations of cost-benefit analysis, nonpaternity, early U.S. experience with sickle cell and Tay-Sachs carrier screening, conditions for overriding patient confidentiality to warn relatives at genetic risk, and ethical rationale for mandatory screening of newborns.


Describes new technologies of testing and possible ethical and social pitfalls of premature distribution.

Examines social, economic, psychological, and ethical issues associated with cystic fibrosis carrier screening in detail, using data from other types of genetic screening. Interdisciplinary and thoroughly researched.


Reconsiders and updates some issues explored by the President’s Commission, as well as new issues posed by multiplex tests and storage of DNA. Emphasizes need for public and professional education, regulation of laboratory quality, and communications understandable to physicians who are not geneticists. Recommends against premature release of new tests and against testing children. Covers newborn screening, informed consent, prenatal diagnosis, and genetic counselling in detail.
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ANNEX 2

INFORMAL LISTING OF
LAWS, REGULATIONS AND GUIDELINES ON
GENETIC SCREENING/TESTING
AND OTHER ASPECTS OF HUMAN GENETICS

INTERNATIONAL TEXTS

COUNCIL OF EUROPE


  Calls for safeguards in gene therapy and in other forms of gene technology.


  Governments of Member States are recommended to adopt legislation in conformity with a series of 14 Principles, or to take other appropriate measures to ensure the implementation of these Principles. An Explanatory Memorandum is appended to the Recommendation.


  Governments of Member States are recommended to be guided in their legislation and policy by a series of Principles and Recommendations. Following an introductory section entitled "Purpose, scope and definitions", a series of 13 Principles are formulated, divided into the following rubrics: I. Rules for good practice in genetic testing and screening; II. Access to genetic tests; III. Data protection and professional secrecy; and IV. Research.

[Further texts to be incorporated]

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1 Prepared by the Health Legislation Unit, WHO, Geneva, Switzerland.
2 The abbreviation IDHL is used throughout for International Digest of Health Legislation, a quarterly journal published by WHO.
EUROPEAN COMMUNITY


Area 5 of the Programme is devoted to "Human genome research". It is specified, inter alia, that "No research modifying, or seeking to modify, the genetic constitution of human beings by alteration of germ cells or of any stage of embryo development which may make these alterations hereditary, will be carried out under this programme".


Stresses the need for cooperation among Member States in this area. Stresses the importance of enhancing "the role of epidemiological research on genetic illnesses in the framework of Community and international research programmes in the fields of medicine and health".


Deals with the conditions under which somatic gene therapy should be undertaken within the European Union; it is stated that "germ line gene therapy on humans is not at the present time ethically acceptable".

NATIONAL TEXTS

AUSTRALIA

National Health and Medical Research Council (NHMRC)


This report includes background information on DNA treatment of patients with inherited diseases (the grounds for gene therapy, DNA treatment of somatic cells, recombinant DNA technology, etc.). Attachments include the following: 1. Medical Research Ethics Committee: terms of reference and membership; 2. NHMRC Statement on human experimentation — list of supplementary notes; and 4. List of Medical Research Ethics Committee's publications.
• **Australia-2.** Guidelines for the Use of Genetic Registers in Medical Research. Issued by the National Health and Medical Research Council in 1991.

These Guidelines deal with: the use of registers in clinical practice (conventions of current use); and the use of genetic registers in medical research (the responsibilities are indicated of, respectively, the keepers of registers and the Institutional Ethics Committee). The following Annexes are appended: A. WHMRC Statement on human experimentation and supplementary notes; and B. WHMRC Statement on scientific practice.

**AUSTRIA**

• **Austria-1.** Ordinance of 20 May 1981 (Serial No. 274) of the Federal Minister for Health and Environmental Protection on priority measures for the maintenance of public health. *(Bundesgesetzblatt für die Republik Österreich, 10 June 1981, No. 106, p. 1389; IDHL, 1982, 33(2), 255-256)*

> Prophylactic measures in the field of human genetics (including genetic counselling, prenatal diagnosis, and cytogenetic examinations) are classed as "priority measures for the preservation of public health". The purpose of these measures is stated to be to reduce the risk of occurrence of genetic diseases. Details are given of the categories of persons covered by these measures.


  Includes a prohibition on interventions involving the germline.

• **Austria-3.** The Gene Technology Law. *(Bundesgesetzblatt für die Republik Österreich, 12 July 1994, No. 158, pp. 4111-4149; IDHL, 1995, 46(1), 000)*

  Imposes a prohibition on the collection and use of genetic data for employment or insurance purposes. Also includes provisions on: genetic testing for medical and research purposes; authorization and compulsory notification of genetic testing activities; genetic counselling; protection of genetic data; gene analysis and somatic cell therapy; and the establishment, composition and functions of the Commission on Gene Technology (its tasks include the preparation of a Gene Technology Code [Gentechnikbuch]).

**BELGIUM**


  Details are given of the Council’s functions, which include promoting the organization of services for genetic counselling.

*Deals essentially with financial aspects.*


*Establishes standards for such services.*


*Establishes detailed standards for such centres, which are required to operate within a university hospital under the jurisdiction of a Belgian university providing complete medical training. Details are given of the functions of the centres.*


*Precludes the provision and use of genetic information in the life insurance context.*

**BRAZIL**


*Among other provisions, prohibits the genetic modification of human germinal cells and in vivo interventions involving human genetic material (except for the treatment of genetic defects).*

**CHINA**


*Includes provisions governing: premarital examinations for the detection of serious genetic diseases; the provision of information to future spouses concerning such diseases; and the qualifications of personnel performing prenatal diagnosis/genetic testing. Sex determination of the fetus is prohibited, except in cases of medical necessity. Physicians are to advise pregnancy termination if the fetus is affected by a serious genetic disease.*
DENMARK

• Denmark-1. Notice of 24 May 1974 of the National Health Service addressed to hospitals, maternity clinics, midwives, public health nurses, medical officers of health, and other physicians in Denmark concerning the examination of neonates for phenylketonuria. (Sundhedsplejen, 1974, No. 4, Den Medicinallovgivning, Suppl., pp. 127-128; IDHL, 1976, 27(1), 90)

Deals with procedural aspects.

• Denmark-2. Notice No. 84 of 21 April 1981 directed to physicians, midwives, and hospitals regarding prophylactic examinations to detect chromosomal diseases, congenital metabolic diseases, neural canal defects, etc. in fetuses. (Ministerialtidende, 1981, 24 July 1981, No. 5, pp. 390-396; IDHL, 1981, 32(4), 701)

Covers prenatal genetic diagnosis, genetic counselling, and testing for genetic disorders.


Includes provisions on certain aspects of gene therapy.

FRANCE

• France-1. Decree No. 88-328 of 8 April 1988 establishing the National Commission on Reproductive Medicine and Biology. (Journal officiel, 9 April 1988, No. 84, pp. 4708-4709)

The Commission includes a Section on Prenatal Diagnosis (one of the members of which must be a specialist in medical genetics).

• France-2. Order of 12 April 1988 determining the clinical laboratories and categories of persons having an exclusive right to carry out prenatal diagnostic procedures. (Journal officiel, 22 April 1988, No. 95, p. 5363; IDHL, 1989, 40(1), 77)

Deals with standards, etc. to be fulfilled in order to perform cytogenetic procedures.


Includes provisions defining conditions under which prenatal diagnosis may be performed in public or private facilities.
• **France-4.** Law No. 94-653 of 29 July 1994 on respect for the human body. *(Journal officiel de la République française, Lois et Décrets, 30 July 1994, No. 175, pp. 11056-11059; IDHL, 1994, 45(4), 498-500)*

> Includes provisions prohibiting eugenics and any alterations to genetic traits with a view to modifying a person’s lineal descent. Also lays down that genetic studies on individual persons may be undertaken only for medical purposes or for scientific research.

• **France-5.** Law No. 94-654 of 29 July 1994 on the donation and use of elements of the human body, medically assisted procreation, and prenatal diagnosis. *(Journal officiel de la République française, Lois et Décrets, 30 July 1994, No. 175, pp. 11060-11068; IDHL, 1994, 45(4), 473-482)*

> Includes detailed provisions on the criteria to be fulfilled by, and requirements governing, prenatal diagnosis (which must be preceded by genetic counselling). Reference is made to the National Commission on Medicine, Reproductive Biology, and Prenatal Diagnosis. Also provides for the insertion of a new Title VI (Predictive medicine and genetic identification) in Book I of the Public Health Code. This includes provisions on the use of genetic finger-printing for medical purposes or for scientific research.

**Avis of the National Ethical Consultative Committee for the Life and Health Sciences**

• **France-6.** Avis N° 5 du 13 mai 1985 sur les problèmes posés par le diagnostic prénatal et périnatal.

• **France-7.** Avis N° 17 du 15 décembre 1989 relatif à la diffusion des techniques d'identification par analyse de l'ADN (techniques des empreintes génétiques).

• **France-8.** Avis N° 22 du 13 décembre 1990 sur la thérapie génique.

• **France-9.** Avis N° 25 du 24 juin 1991 sur l'application des tests génétiques aux études individuelles, études familiales et études de population (problèmes des "banques" de l'ADN, des "banques" de cellules et de l'informatisation des données).


**GERMANY**


> Includes provisions proscribing germline gene therapy.

**Lower Saxony**

• **Germany (Lower Saxony)-1.** Circular of 6 July 1978 on prophylactic examinations
of neonates. Early diagnosis of galactosaemia and hypothyroidism. (IDHL, 1980, 31(3), 529)

Deals with standards for criteria governing the examinations concerned.

INDIA


This Act includes the following Chapters: II. Regulation of genetic counselling centres, genetic laboratories and genetic clinics; III. Regulation of pre-natal diagnostic techniques; and VI. Registration of genetic counselling centres, genetic laboratories and genetic clinics. According to The Times of India (27 July 1994, p. 8), this Act has received the assent of both Houses of the Indian Legislature.

Rajasthan


This Law regulates the use of "pre-natal diagnostic techniques for the purpose of detecting genetic or metabolic disorders or chromosomal abnormalities of certain congenital malformations or sex-linked disorders and for the prevention of the misuse of such techniques for the purpose of pre-natal sex determination leading to female foeticide".

ITALY

Friuli-Venezia Giulia


Includes provisions on genetic counselling before conception and prenatal diagnosis in order to detect and prevent diseases of the embryo and fetus due to maternal infection and the genetic causes of diseases and malformations in the mother and child.

NETHERLANDS


This Law evidently applies to genetic screening (although no reference is made thereto in the Law).
NORWAY


  Includes detailed provisions on: genetic counselling before preimplantation diagnosis; genetic counselling in the context of prenatal diagnosis; postnatal genetic testing; restrictions on the use of genetic information; gene therapy; and various other aspects of genetic testing and counselling.

POLAND

- Poland-1. Law of 7 January 1993 on family planning, protection of human fetuses, and the conditions under which pregnancy termination is permissible. (IDHL, 1993, 44(2), 253-255)

  An amendment to the Penal Code establishes criteria for "prenatal examinations that do not significantly increase the risk of abortion". Thus, such examinations are authorized in cases in which, inter alia: (1) the unborn child belongs to a family manifesting a genetic "burden"; (2) it is assumed that a genetic condition may be cured or that it is possible to remedy that condition or to limit its effects at the fetal stage; and (3) a presumption exists that the fetus presents a serious defect.

PORTUGAL


  The functions of the Institute are defined.


  Family planning services are to include genetic counselling.

SOUTH AFRICA


  Includes provisions intended to ensure that genetic factors are taken into account in the context of artificial insemination procedures. Thus, it is laid down, inter alia, that, where the recipient or donor comes from a population group "in which the individual runs a high risk of being a carrier of a specific genetic defect, for example Tay-Sachs disease or thalassemia, the recipient and the donor are tested for the characteristics concerned and that the gamete of a donor with the same characteristics as the gamete of the recipient is not used for artificial insemination" (there are similar provisions that apply in other contexts in which a genetic risk exists).
SPAIN


*Includes provisions aimed at the development of genetic counselling and testing of couples, on a voluntary basis.*


*Provides for the establishment of a network of laboratories providing genetic counselling.*


*Funds are identified for a "Metabolic-Genetic Group" as part of the arrangements for the implementation of the Plan.*


*Includes provisions enabling the use of genetic technology for in vitro and in vivo prenatal diagnosis of genetic/hereditary diseases.*

Castile-La Mancha


*Establishes a Programme for the Early Detection of Metabolic Disorders.*

Catalonia


*Includes provisions dealing with genetic/hereditary diseases that can be diagnosed by pre-implantation techniques.*
Valencia


Includes provisions on genetic counselling.

SWEDEN


Special authorizations are required for investigations of the human genetic patrimony involving analysis of gene DNA or RNA, where such investigations constitute or form part of a general health examination.


Experimentation on fertilized oocytes for research or therapeutic purposes must not have as its objective the development of "methods aimed at causing heritable genetic effects".

SWITZERLAND


Amends the Federal Constitution of 29 May 1874 by the insertion of a new Sec. 24 novies. This Section lays down that the Confederation is to issue provisions concerning the use of the germ-cell and the "human genetic patrimony". The following are among the principles that are to govern such provisions: interventions affecting the genetic heritage of human gametes and embryos shall not be admissible; the germ-cell and genetic heritage of non-human species may not be transferred to the human germ-cell heritage, nor fused with it; the human germ-cell heritage and products derived from embryos may not be the subject of commerce; and the genetic heritage of a person may only be analysed, recorded, and disclosed with that person's consent, or on the basis of a legal provision.

Swiss Academy of Medical Sciences

The Guidelines are subdivided into the following rubrics: 1. Scope; 2. Medical indications for genetic investigations; and 3. Recommendations for the performance of medical-genetic investigations. Also included are a Preamble and a Commentary, as well as discussions on prenatal diagnosis, counselling, and ethical issues.

Canton of Basel-Land


Includes provisions laying down that interventions on the genetic material of human cells are prohibited.

Canton of Geneva

• Switzerland (Geneva)-1. Regulations of 13 April 1994 concerning clinical research involving interventions in the field of human genetic manipulation. (Feuille d'Avis Officielle, 20 April 1994, No. 44, p. 1; IDHL, 1994, 44(3), 333-334)

Detailed standards are laid down for certain categories of clinical research in the field of human genetics.

UNITED KINGDOM

Department of Health


This non-statutory advisory body was set up further to a recommendation by the Committee on the Ethics of Gene Therapy, chaired by Sir Cecil Clothier. Its terms of reference are as follows: (a) to consider and advise on the acceptability of proposals for gene therapy research on human subjects, on ethical grounds, taking account of the scientific merits of the proposals and the potential benefits and risks; (b) to work with other agencies which have responsibilities in this field, including local research ethics committees and agencies which have statutory responsibilities — the Medicines Control Agency, the Health and Safety Executive, and the Department of the Environment; and (c) to provide advice to UK Health Ministers on developments in gene therapy research and their implications.

Medicines Control Agency


[To be completed when final version available]
Committee on the Ethics of Gene Therapy


  This report includes, in addition to an introductory guide to genes, genetic disorders, and gene therapy, sections on: the official basis of medical practice, research, and gene therapy; somatic cell gene therapy; germline gene therapy; the supervision of gene therapy; and recommendations and conclusions.

Nuffield Council on Bioethics


  The Conclusions of this report are subdivided into the following rubrics: 1. Consent; II. Confidentiality; III. Employment; IV. Insurance; V. Public policy; and VI. Implementation of screening programmes.

UNITED STATES OF AMERICA


  Amends the Public Health Service Act by, inter alia, the insertion of a new Subpart 3 (National Center for Human Genome Research) in Part E of Title IV. The Center's "general purpose" is to "characterize the structure and function of the human genome, including the mapping and sequencing of individual genes", including: planning and coordinating the research goal of the Human Genome Project (HGP); coordinating international genome research; communicating advances in genome science to the public; and reviewing proposals to address the ethical and legal issues associated with the HGP (including legal issues regarding patents).


  These Guidelines supersede all earlier versions, their purpose being to specify practices for constructing and handling: (i) recombinant deoxyribonucleic acid (DNA) molecules; and (ii) organisms and viruses containing recombinant DNA molecules.

  Certain of the provisions of the Guidelines deal with the transfer of recombinant DNA, etc., into one or more human subjects. Specific reference should be made to a highly detailed Appendix M (Points to consider in the design and submission of protocols for the transfer of recombinant DNA molecules into the genome of one or more human subjects). Included in this Appendix are rubrics dealing with: research design, and anticipated risks and benefits; selection of patients; informed consent; and privacy and confidentiality.


California

• United States of America (California)-1. An Act (Chapter 761) to amend, repeal, and add Section 1374.7 of the Health and Safety Code, and to amend, repeal, and add Sections 10123.3, 10140, and 11512.95 of, and to add Article 2.6 (commencing with Section 10146) to Chapter 1 of Part 2 of Division 2 of, the Insurance Code, relating to discrimination. Date of enactment: 23 September 1994. (IDHL, 1995, 46(2), 000-000)

This Law prohibits health insurance companies from discriminating against policy-holders or applicants for health insurance policies on the basis of asymptomatic genetic characteristics.

IN PREPARATION

• Council of Europe. Draft Bioethics Convention.

The "declassified" draft text (July 1994) includes three Articles dealing with human genetics, viz.: 16 (Human genome), 17 (Tests predictive of genetic disease), and 18 (Communication of results) (the latter Article deals with the "communication of results of genetic testing outside the health field").

• UNESCO (International Bioethics Committee). Draft Declaration on the Protection of the Human Genome.

The available draft (dated 7 March 1995) comprises rubrics addressing the following: (a) Aims of research on the human genome; (b) Operations affecting the human genome and human rights and freedoms; (c) Rights and obligations of researchers; (d) Duties and responsibilities towards others; (e) International co-operation; and (f) Implementation of the Declaration.

* * * *