


Ethical issues in medical genetics and genetic services

WHO documents: 1999, 2003


Summarised by Hanan Hamamy

Training Course in Sexual and Reproductive Health Research
Geneva 2010



Relevant Ethical Principles in Medicine

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




Ethical Principles Applied to Genetics Services (1 of 3)

1. Fair allocation of public resources to those who most need them (justice).
2. Freedom of choice in all matters relevant to genetics. The woman should be the final decision maker in reproductive choices (autonomy).
3. Voluntary approach necessary in services, including approaches to testing and treatment; avoid coercion by government, society, or health professionals (autonomy).
4. Respect for human diversity and for those whose views are in the minority (autonomy, non- maleficence).
5. Respect for people's basic intelligence, regardless of their knowledge (autonomy).




Ethical Principles Applied to Genetics Services (2 of 3)

6. Education about genetics for the public, medical and other health professionals, teachers, clergy, and other persons who are sources of religious information (beneficence).
7. Close cooperation with patient and parent organizations, if such organizations exist (autonomy).
8. Prevention of unfair discrimination or favouritism in employment, insurance, or schooling based on genetic information (non-maleficence).
9. Teamwork with other professionals through a network of referrals. When possible, help individuals and families become informed members of the team (beneficence, autonomy).



Ethical Principles Applied to Genetics Services (3 of 3)

10. Use of non-discriminatory language that respects individuals as persons (autonomy).
11. Timely provision of indicated services or follow-up treatment (non-maleficence).
12. Refraining from providing tests or procedures not medically indicated (non-maleficence).
13. Providing ongoing quality control of services, including laboratory procedures (non-maleficence).




Education is a Key to Ethical Genetics Services

Education of the public

- Revising school curricula
- Scientifically based media messages

Professional Education

- Revising medical and nursing schools curricula
- Training of health care providers and policymakers



The objectives of medical genetics education may be fulfilled if health care providers can acquire the following:

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



Ethical Principles Applied to Genetic Counselling (1 of 2)

1. Respect for persons and families, including full disclosure, respect for people's decisions, accurate and unbiased information (autonomy).
2. Preservation of family integrity (autonomy, non-maleficence).
3. Full disclosure to individuals and families of all information relevant to health (non-maleficence, autonomy).
4. Protection of the privacy of individuals and families from unjustified intrusions by employers, insurers, and schools (non-maleficence).
5. Information to individuals and families about possible misuses of genetic information by institutional third parties (non-maleficence).
6. Informing individuals that it is the individual's ethical duty to tell blood relatives that the relatives may be at genetic risk (non-maleficence).



Ethical Principles Applied to Genetic Counselling (2 of 2)

7. Informing individuals about the wisdom of disclosing their carrier status to spouse/partner if children are intended, and the possibility of harmful effects on the marriage from disclosure (nonmaleficence).
8. Informing people of their moral duties to disclose a genetic status that may affect public safety (non-maleficence).
9. Unbiased presentation of information, insofar as this is possible (autonomy).
10. Non-directive approach, except when treatment is available (autonomy, beneficence).
11. Children and adolescents to be involved in decisions affecting them, whenever possible (autonomy).
12. Duty to recontact if appropriate and desired (non-maleficence, beneficence, autonomy).

Premarital counseling

In cultures where arranged marriages are the norm, premarital testing for recessive disorders common in particular populations may avert unions at high genetic risk. In order to prevent stigmatization of individuals or families, it is important that test results be kept strictly confidential.

Each individual involved (that is, the members of the prospective couple) should have full knowledge of the test results, together with full education and supportive counselling.

It may be necessary to educate other family members in order to prevent prejudice against carriers.



Proposed Ethical Guidelines for Genetic Screening and Testing (1 of 2)

1. Genetic screening and testing should be voluntary (autonomy); not mandatory, with the exception noted in the last point below.
2. Genetic screening and testing should be preceded by adequate information about the purpose and possible outcomes of the screen or test and potential choices to be made (autonomy, nonmaleficence).
3. Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened (autonomy).
4. Results should not be disclosed to employers, insurers, or others without the individual's consent, in order to avoid possible discrimination (autonomy, non-maleficence).



Proposed Ethical Guidelines for Genetic Screening and Testing (2 of 2)

5. In rare cases where disclosure may be in the best interests of the individual or of public safety, the health provider may work with the individual towards a decision by him or her (beneficence, nonmaleficence, justice).
6. Test results should be followed by genetic counselling, particularly when they indicate the presence of a mutation or a genetic condition (autonomy, beneficence).
7. If treatment or prevention exists or is available, this should be offered with a minimum of delay (beneficence, non-maleficence).
8. Newborn screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn (beneficence, justice).

Autonomy and Informed Consent (1 of 2)

a) **Applicable to clinical practice:**

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

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

Autonomy and Informed Consent (1 of 2)

b) Applicable to Research and Quality Control:


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

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



Proposed Guidelines for Presymptomatic and Susceptibility Testing

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



Disclosure and Confidentiality

(1 of 3)

1. Professionals should disclose to tested individuals all test results relevant to their health or the health of a fetus. Adequate information is a prerequisite for free choice and is necessary to the open communication and trust that should mark the relationship between the provider and the person counselled.
2. Test results, including normal results, should be communicated to the tested person without undue delay.
3. Test results not directly relevant to health, such as non-paternity, or the sex of the fetus in the absence of an X-linked disorder, may be withheld if this appears necessary to protect a vulnerable party or if prescribed by national law.



Disclosure and Confidentiality

(2 of 3)

4. The wishes of individuals and families not to know genetic information, including test results, should be respected, except in testing of newborn babies or children for treatable conditions.
5. Information that could cause grave psychological or social harm may be temporarily withheld. Within the general duty of disclosure, the counsellor may exercise judgement about when a tested person is ready to receive information.
6. If a couple intends to have children, individuals should be encouraged to share genetic information with their partners.
7. Where appropriate, as part of their general duty to educate, counsellors should inform people that genetic information may be useful to their relatives and may invite individuals to ask the relatives to seek genetic counselling.


Disclosure and Confidentiality

(3 of 3)

8. The provision of genetic information to relatives about the family so as to learn their own genetic risks should be possible, especially when harm can be avoided.
9. Results of carrier tests, presymptomatic tests, susceptibility tests, and prenatal tests should be kept confidential from employers, health insurers, schools and government agencies. People should not be penalized or rewarded for their genetic constitutions. Information about a symptomatic condition may be disclosed as part of general medical information, in accordance with laws and practices in different countries.
10. Registries (if any) should be protected by the strictest standards of confidentiality.


Proposed Ethical Guidelines for Prenatal Diagnosis (1 of 2)

1. Equitable distribution of genetics services, including prenatal diagnosis, is owed first to those with the greatest medical need, regardless of ability to pay, or any other considerations (justice).
2. Prenatal diagnosis should be voluntary in nature. The prospective parents should decide whether a genetic disorder warrants prenatal diagnosis or termination of a pregnancy with an affected fetus (autonomy).
3. If prenatal diagnosis is medically indicated, it should be available regardless of a couple's stated views on abortion. Prenatal diagnosis may, in some cases, be used to prepare for the birth of a child with a disorder (autonomy). Prenatal diagnosis for adult-onset disorders may require special counselling , so as to avoid testing of children who may be carried to term.
4. Prenatal diagnosis is done only to give parents and physicians information about the health of the fetus. The use of prenatal diagnosis for gender selection, apart from a situation with risk for sexlinked disorders, is not acceptable (non-maleficence). Prenatal diagnosis for paternity testing, except in cases of rape or incest, is generally unacceptable, but should be considered on a case-by-case basis.



Proposed Ethical Guidelines for Prenatal Diagnosis (2 of 2)


5. Prenatal diagnosis solely for relief of maternal anxiety, in the absence of medical indications, should have lower priority in allocation of resources than prenatal diagnosis with medical indications (justice).
6. Counselling should precede prenatal diagnosis (non-maleficence).
7. Physicians should disclose all clinically relevant findings to the woman or couple, including the full range of variability in the manifestations of the condition under discussion (autonomy).
8. The woman's and/or couple's choices in a pregnancy with an affected fetus should be respected and protected, within the framework of the family and of the laws, culture and social structure of the country. The couple, not the health professional, should make the choice (autonomy).



Proposed Guidelines for Counselling prior to Prenatal Diagnosis (1 of 3)


Counselling should include the following points as a minimum:

1. Name(s) and general characteristics of the major disorder(s) that the test may identify. The list of disorders need not be exhaustive. The characteristics of the disorder(s) should be described also in terms of their effects on the future child, on the parents, and on family life.
2. Possibilities for treatment of the disorder(s) after birth and availability of supportive care.
3. Description of the likelihood (risk) that the fetus may have the disorder(s). Risks should be expressed in several ways (as a percent, as a proportion, and verbally).
4. The possibility of unfavourable test results or of fortuitous or unexpected findings.




Proposed Guidelines for Counselling prior to Prenatal Diagnosis (2 of 3)

5. Alternatives available for those with an affected fetus, for example, carrying the fetus to term and caring for the child at home; placing the child in an institutional setting, if available; placing the child for adoption; termination of pregnancy; prenatal treatment for the fetus or early treatment after birth.
6. The possibility of ambiguous laboratory or ultrasonography results.
7. Information that, because most conditions diagnosed in the fetus cannot be treated before birth, knowing about the existence of a condition may not help the fetus.
8. Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth, or are not tested for unless the family is known to be at high risk.




Proposed Guidelines for Counselling prior to Prenatal Diagnosis (3 of 3)

9. The medical risks to fetus and mother posed by the testing procedure.
10. Non-medical risks, if any (e.g., to parental employment or health care, where applicable).
11. Information that non-invasive screens used early in pregnancy, such as maternal serum alfafetoprotein (MSAFP), may be the first step on the road to prenatal diagnosis and a possible decision about abortion.
12. Costs of the test and sources of reimbursement for the mother or couple, if applicable.
13. Names and addresses of genetic support groups or organizations for persons with genetic disorders, that people can contact if they wish.




Why termination of a pregnancy with an affected fetus is difficult

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



Proposed Ethical Guidelines for Access to Banked DNA (1 of 2)

1. A blanket informed consent that would allow use of a sample in future projects is the most efficient approach.
2. Control of DNA may be familial, not only individual. Blood relatives may have access to stored DNA for purposes of learning their own genetic status, but not for purposes of learning the donor's status.
3. Family members should have access regardless of whether they contributed financially to the banking of the DNA.
4. DNA should be stored as long as it could be of benefit to living or future relatives or fetuses.
5. Attempts should be made to inform families, at regular intervals, of new developments in testing and treatment. Donors should inform DNA banks of current addresses for follow-up.



Proposed Ethical Guidelines for Access to Banked DNA (2 of 2)

6. After all relatives have died or all attempts to contact survivors have failed, DNA may be destroyed.
7. Spouses should not have access to DNA banks without the donor's consent, but may be informed that DNA has been banked. If the couple is considering having children, it is the moral obligation of the party whose DNA has been banked to provide the spouse any relevant information.
8. Except for forensic purposes or instances when the information is directly relevant to public safety, there should be no access for institutions without the donor's consent. Insurance companies, employers, schools, government agencies, and other institutional third parties that may be able to coerce consent should not be allowed access, even with the individual's consent.
9. Qualified researchers should have access if identifying characteristics are removed.
10. Potentially valuable specimens that could be useful to concerned families in the future should be saved and should be available.



Review of Ethical Issues (1 of 3)

1. Existing genetics services in a nation should be available equally to everyone regardless of ability to pay and should be provided first to those whose need is greatest.
2. Genetic counselling should be non-directive.
3. All genetics services, including screening, counselling, and testing, should be voluntary, with the exception of screening newborns for conditions for which early and available treatment would benefit the newborn.
4. All clinically relevant information that may affect the health of an individual or fetus should be disclosed.

Review of Ethical Issues (2 of 3)

5. Confidentiality of genetic information should be maintained. When there is a high risk of serious harm to family members at genetic risk, the information should be used to avert this harm. If the individual refuses to tell her/his family, the professional may consider overriding confidentiality.
6. Individual privacy should be protected from institutional third parties, such as employers, insurers, schools, commercial entities, and government agencies.
7. Prenatal diagnosis should be performed only for reasons relevant to the health of the fetus and only to detect genetic conditions or fetal malformations.
8. Choices relevant to genetics services, including choices about counselling, screening, testing, contraception, assisted procreation where culturally accepted, and abortion following prenatal diagnosis, where legal, should be available on a voluntary basis and should be respected.



Review of Ethical Issues (3 of 3)

9. Optimum support and education should be provided for children and families with genetic conditions.
10. Adopted children and others with biological relationships outside the family should be able to receive information about their biological relatives, under strict anonymity rules.
11. Research protocols should follow established procedures for review and informed consent.
12. Protocols for experimental human gene therapy should receive national review, with attention to the potential benefits or risks arising from various approaches to therapy.