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"GenEthics":
Ethical Issues in Medical Genetics

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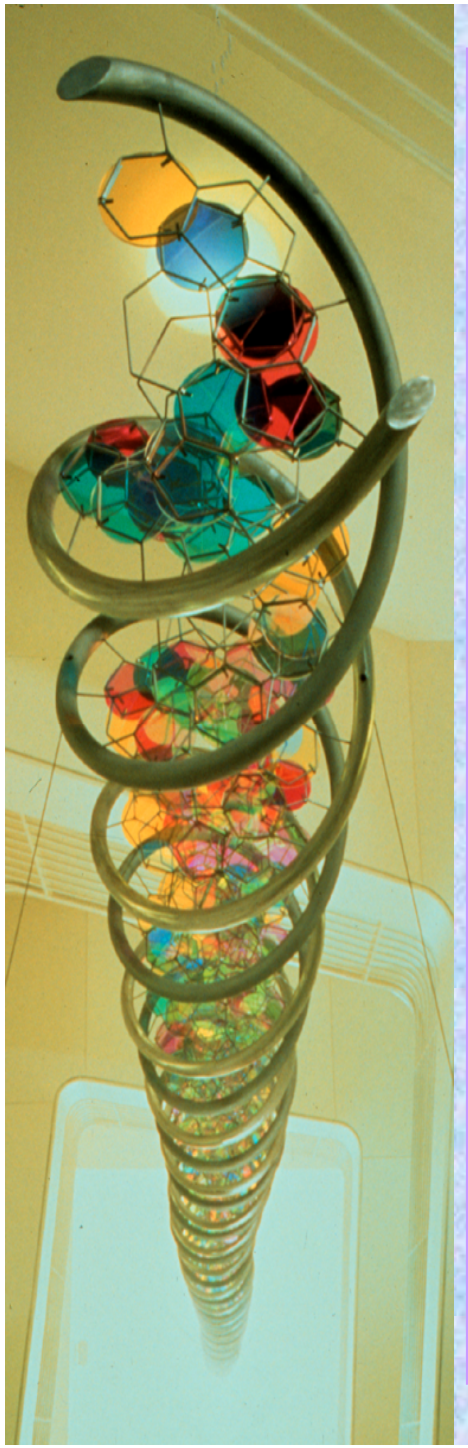

Importance of Genetics in Medicine

- Hereditary conditions affect millions of families throughout the world.
- About 5% of all pregnancies result in the birth of a child with a significant genetic disorder, congenital malformation or disability.
- An estimated 43% of cases of severe mental retardation (IQ <50) are caused by single genes or chromosomal abnormalities (Institute of Medicine, 1994).
- In developed nations, totally or partially hereditary conditions account for about 36% to 53% of paediatric hospital admissions (Institute of Medicine, 1994).
- In developing nations hereditary conditions account for about 15% to 25% of perinatal and infant mortality (Verma and Singh, 1989; Penchaszadeh, 1993 a,b).
- Most non-infectious diseases, which are the major causes of death in developed nations, may have a genetic component (Holtzman, 1989).

Resources for Addressing

Ethical Issues in Medical Genetics

"Ethics" is a generic term for various ways of understanding and examining the moral life and for resolving ethical problems (Beauchamp & Childress, 1994).

Ethics, as a field in Philosophy or Religion, is concerned with systematic reflection on the moral life and its conflicts.

Biomedical Ethics (or bioethics) is an interdisciplinary field for the systematic study of ethical issues that arise in research, medicine and society (WHO 1992a; UNESCO, 1993).

Resources for Ethical Guidance

Approaches to Medical Ethics are varied.

With regarding principles-based ethics: *Ethical Principles in Medicine*

The traditional sources of ethical guidelines in medicine apply also to medical genetics, which is a field of 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 issues can be identified within four large arenas:

(1) Research and its application to all forms of life, from bacteria and viruses to plants, animals, and humans;

(2) Allocation and delivery of health care resources;

(3) Ethical problems that arise in clinical encounters between health care professionals and patients,

(4) Ethical problems in preventive medicine and public health.

Genetic Vs. Medical Data

Medical Genetics' main concerns, however, extend beyond those of the traditional structure of medicine and the physician-patient relationship.

For example:

a) Genetic information may affect an entire family, rather than only the individual (ability of the data to be generalised);

b) Genetic information may have predictive potentials in development of a particular trait or disease in future in addition to its diagnostic and prognostic application;

c) Genetic information have permanent value and validity throughout the individual's life;

d) Genetic information and the choices of the present may affect future generations.

Ethical Principles Applied to Genetics Services

1. Fair allocation of public resources to those who most need them (justice).
2. Freedom of choice in all matters relevant to genetics. The woman should be the final decision-maker in reproductive choices (autonomy).
3. Voluntary approach necessary in services, including approaches to testing and treatment; avoid coercion by government, society, or health professionals (autonomy).
4. Respect for human diversity and for those whose views are in the minority (autonomy, non-maleficence).
5. Respect for people's basic intelligence, regardless of their knowledge (autonomy).
6. Education about genetics for the public, medical and other health professionals, teachers, clergy, and other persons who are sources of religious information (beneficence).
7. Prevention of unfair discrimination or favouritism in employment, insurance, or schooling based on genetic information (non-maleficence).
8. Refraining from providing tests or procedures not medically indicated (non-maleficence).
9. Providing ongoing quality control of services, including laboratory procedures (non-maleficence).

Education as the Key to Ethical Genetics Services

Public Education:

The goals of medical genetics can be optimally fulfilled only in the context of an educated, informed public.

Education about human reproduction and genetics should be part of the educational heritage of every person (Bankowski and Capron, 1991; Fujiki et al, 1991).

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

In the long run, genetics education for the public can best be achieved through education in schools.

Ethical Principles Applied to Genetic Counselling

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

Essential goals of genetic counselling

(1) Helping individuals/couples understand their options and the present state of medical knowledge so they can make informed decisions;

(2) Helping individuals/couples adjust to and cope with their genetic problems;

(3) The removal or lessening of guilt or anxiety;

(4) Helping individuals/couples achieve their parenting goals

Proposed Ethical Guidelines for Genetic Screening & Testing

1. Genetic screening and testing should be voluntary (autonomy); not mandatory, with one exception: Newborn screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn (beneficence, justice).
2. Genetic screening and testing should be preceded by adequate information about the purpose and possible outcomes of the screen or test and potential choices to be made (autonomy, non-maleficence).
3. Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened (autonomy).
4. Results should not be disclosed to employers, insurers, or others without the individual's consent, in order to avoid possible discrimination (autonomy, non-maleficence).
5. In rare cases where disclosure may be in the best interests of the individual or of public safety, the health provider may work with the individual towards a decision by him or her (beneficence, 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).

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).

Proposed Ethical Guidelines for Access to Banked DNA

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

Proposed Ethical Guidelines for Prenatal Diagnosis

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

Proposed Guidelines for Counselling prior to Prenatal Diagnosis

Counselling should include the following points as a minimum:

1. Name(s) and general characteristics of the major disorder(s) that the test may identify. The list of disorders need not be exhaustive. The characteristics of the disorder(s) should be described also in terms of their effects on the future child, on the parents, and on family life.
2. Possibilities for treatment of the disorder(s) after birth and availability of supportive care.
3. Description of the likelihood (risk) that the fetus may have the disorder(s). Risks should be expressed in several ways (as a percent, as a proportion, and verbally).
4. The possibility of unfavourable test results or of fortuitous or unexpected findings.
5. Alternatives available for those with an affected fetus, for example, carrying the fetus to term and caring for the child at home; placing the child in an institutional setting, if available; placing the child for adoption; termination of pregnancy; prenatal treatment for the fetus or early treatment after birth.
6. The possibility of ambiguous laboratory or ultrasonography results.
7. Information that, because most conditions diagnosed in the fetus cannot be treated before birth, knowing about the existence of a condition may not help the fetus.
8. Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth, or are not tested for unless the family is known to be at high risk.
9. The medical risks to fetus and mother posed by the testing procedure.
10. Non-medical risks, if any (e.g., to parental employment or health care, where applicable).
11. Information that non-invasive screens used early in pregnancy, such as maternal serum alphafetoprotein (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.

"Is Medical Genetics Eugenics?"

Meanings of the term "Eugenics": The word "eugenics", from the Greek for "good birth", was coined in 1883 by Sir Francis Galton.

Although the term still carries its original Galtonian meaning of "healthy birth" in some parts of the world, it is usually employed as a pejorative today, without careful attention to its meaning. Genetics aims to improve the lives of individuals and families (but not to "improve" the genetic health of the society), that in human populations there are no "superior" or inferior" genomes, and that human diversity contributes to the survival and richness of humanity.

Eugenics: "A coercive policy intended to further a reproductive goal, against the rights, freedoms, and choices of the individual." For purposes of this definition, "coercion" includes laws, regulations, positive or negative incentives (including lack of accessibility to affordable medical services)

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

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

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

Disclosure and Confidentiality

1. Professionals should disclose to tested individuals all test results relevant to their health or the health of a fetus. Adequate information is a prerequisite for free choice and is necessary to the open communication and trust that should mark the relationship between the provider and the person counselled.
2. Test results, including normal results, should be communicated to the tested person without undue delay.
3. Test results not directly relevant to health, such as non-paternity, or the sex of the fetus in the absence of an X-linked disorder, may be withheld if this appears necessary to protect a vulnerable party or if prescribed by national law.
4. The wishes of individuals and families not to know genetic information, including test results, should be respected, except in testing of newborn babies or children for treatable conditions.
5. Information that could cause grave psychological or social harm may be temporarily withheld.

Within the general duty of disclosure, the counsellor may exercise judgement about when a tested person is ready to receive information.

6. If a couple intends to have children, individuals should be encouraged to share genetic information with their partners.
7. Where appropriate, as part of their general duty to educate, counsellors should inform people that genetic information may be useful to their relatives and may invite individuals to ask the relatives to seek genetic counselling.
8. The provision of genetic information to relatives about the family so as to learn their own genetic risks should be possible, especially when harm can be avoided.
9. Results of carrier tests, presymptomatic tests, susceptibility tests, and prenatal tests should be kept confidential from employers, health insurers, schools and government agencies. People should not be penalized or rewarded for their genetic constitutions. Information about a symptomatic condition may be disclosed as part of general medical information, in accordance with laws and practices in different countries.
10. Registries (if any) should be protected by the strictest standards of confidentiality.

Review Of Ethical Issues

1. Existing genetics services in a nation should be available equally to everyone regardless of ability to pay and should be provided first to those whose need is greatest.
2. Genetic counselling should be non-directive.
3. All genetics services, including screening, counselling, and testing, should be voluntary, with the exception of screening newborns for conditions for which early and available treatment would benefit the newborn.
4. All clinically relevant information that may affect the health of an individual or fetus should be disclosed.
5. Confidentiality of genetic information should be maintained. When there is a high risk of serious harm to family members at genetic risk, the information should be used to avert this harm. If the individual refuses to tell her/his family, the professional may consider overriding confidentiality.
6. Individual privacy should be protected from institutional third parties, such as employers, insurers, schools, commercial entities, and government agencies.
7. Prenatal diagnosis should be performed only for reasons relevant to the health of the fetus and only to detect genetic conditions or fetal malformations.
8. Choices relevant to genetics services, including choices about counselling, screening, testing, contraception, assisted procreation where culturally accepted, and abortion following prenatal diagnosis, where legal, should be available on a voluntary basis and should be respected.
9. Optimum support and education should be provided for children and families with genetic conditions.
10. Adopted children and others with biological relationships outside the family should be able to receive information about their biological relatives, under strict anonymity rules.
11. Research protocols should follow established procedures for review and informed consent.
12. Protocols for experimental human gene therapy should receive national review, with attention to the potential benefits or risks arising from various approaches to therapy.


Thank you!