

GUIDELINE OF THE MALAYSIAN MEDICAL COUNCIL

MMC GUIDELINE 010/2006

MEDICAL GENETICS AND GENETIC SERVICES



Malaysian Medical Council

PRELUDE

This Guideline complements, and should be read in conjunction with the Code of Professional Conduct and Guidelines issued by the Malaysian Medical Council (MMC) and other related Guidelines.

In this Guidelines, the words “doctor”, “physician”, “medical practitioner” and “practitioner” are used interchangeably, and refer to any person registered as a medical practitioner under the Medical Act 1971. The words “hospital” and “healthcare facility and service” are used interchangeably and refer to any premises in which members of the public receive healthcare services. Words denoting one gender shall include the other gender. Words denoting a singular number shall include the plural and vice versa.

Adopted by the Malaysian Medical Council on 14 November 2006

FOREWORD

The Malaysian Medical Council, with the objective of ensuring that registered medical practitioners are fully aware of the codes of professional medical practice, issues directives and guidelines from time to time. The purpose of these codes, guidelines and directives is to safeguard the patient and members of the public, to ensure propriety in professional practice and to prevent abuse of professional privileges.

The Guideline are designed to complement, and should be read in conjunction with, the Medical Act Regulations, Code of Professional Conduct of the Malaysian Medical Council and other Guidelines issued by the Council or any related organisation, as well as any statute or statutory provisions in force and all related statutory instruments or orders made pursuant thereto.

This Guideline on the **Medical Genetics and Genetic Services** has been prepared with careful attention to detail, cognisant of the current international stand on the subject. The draft has been reviewed numerous times by the Malaysian Medical Council and includes valuable response from individuals, organisations and professional bodies in the country, before formal adoption by the Council.

The Guideline is available in the printed form as well as in the MMC website. Registered medical practitioners are advised to familiarise themselves with the contents, as they will serve as documents to refer to or to seek clarifications from, when they need guidance on matters of professional ethics, codes of professional conduct and medical practice in general.

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MEDICAL GENETICS & GENETIC SERVICES

SUMMARY

The advances in human genetics during the last twenty years have revolutionized knowledge of the role of inheritance in health and disease.

These advances will only be acceptable if their application is carried out with due consideration of accepted codes of medical ethics and social ethics, in relation to autonomy, justice and education and to the laws of the country.

Ethical consideration in genetic services must favour the population's quest to benefit with predictable safety from the application of the advances of genetic knowledge.

This Guideline on Medical Genetics and Genetic Services addresses various issues from counseling and consent for genetic screening, genetic registers, prenatal diagnosis, as well as other relevant related aspects.

Aspects of DNA banking as well as the role of DNA profiling in diagnosis and treatment of diseases, insurance, medico-legal and paternity disputes and in criminal investigative procedures are covered.

1. INTRODUCTION

The advances in human genetics that have occurred during the past twenty years have revolutionized the knowledge of the role of inheritance in health and disease. We now know that our DNA determines not only the cause of catastrophic single-gene disorders, which affect millions of persons worldwide, but also predisposition to cancer, heart disease, psychiatric disorders and even to some infectious diseases. However, these advances will only be acceptable if their application is carried out ethically, with regard to autonomy, justice, education and their beliefs and the laws of each nation.

In Malaysia, the genetic service is at infancy. It is therefore timely that ethical consideration in genetic service should be given due regard so that the population can benefit from the advances of genetic knowledge from the beginning and without any harm.

The general principles of medical genetics outlined below are modified from the recommendations of WHO and have been adopted by many countries in the world.

In general, it is ethically imperative that genetic data should only be used to the advantage of members of family or ethnic group, and never to stigmatize or discriminate against them.

There shall be no compulsory genetic testing of adult individuals or populations. Every test shall be offered in such a way that individuals and families are free to refuse or accept according to their wishes and moral beliefs. All testing should be preceded by adequate information about the purpose and possible outcomes of the test and potential choices that may arise. Children shall only be tested when it is for the purpose of better medical care, as in the case of newborn screening when early treatment will be of benefit to the child.

Prenatal diagnosis should be offered to those who need it, but there must be no pressure on couples to accept such testing, nor to use the results of the test to compel either continuing or terminating the pregnancy when the fetus is affected with a genetic disorder.

Decision in the context of reproduction should rest with those being tested, not with physicians or the government.

The woman should be an important decision-maker in all matters related to reproduction.

Prenatal diagnosis should be done only to give parents and physicians information about the health of the foetus; its use for paternity testing, except in cases of rape or incest, or for gender selection, apart from sex-linked disorders, is not acceptable.

Genetic services for the prevention, diagnosis and treatment of disease should be available to all, without regard to ability to pay, and should be provided first to those whose needs are greatest.

Genetic data should only be used to advantage and empower an individual or family, and for better treatment or prevention of disease. Data relevant to health care should be collected and kept by medical geneticists in secure confidential records.

Genetic data should not be given out to insurance companies, employers, schools or government, except with the full informed consent of the person tested. In some countries it may be possible, or necessary, to protect both confidentiality and non-discrimination through legal means.

Genetic counseling is the provision of accurate, full and unbiased information in a caring, professional relationship that offers guidance, but allows individuals and families to come to their own decisions.

Counseling is essential before any genetic testing is carried out, and should continue afterwards if the results entail choices for the person and family tested.

Genetic counseling should be available to all, and should be as non-directive as possible.

Education about genetics for the public and health care professionals is of paramount importance.

2. ETHICAL PRINCIPLES IN MEDICAL GENETICS.

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

Relevant Ethical Principles in Medical Practice

- Respect for the autonomy of persons: respecting the self-determination of individuals and protecting those 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 and maximizing benefits to their health and minimising harm;

- Justice: treating persons with fairness and equity, and distributing the benefits and burdens of health care as fairly as possible in society.

3. GOALS AND PRACTICES OF MEDICAL GENETICS.

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

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

4. APPLICATION OF ETHICAL PRINCIPLES TO GENETIC SERVICES

Ethical Principles Applied to Genetic Services

- Fair allocation of public resources to those who most need them (justice).

- Freedom of choice in all matters relevant to genetics. The woman should be an important decision-maker in reproductive matters (autonomy).
- Voluntary approach necessary in services, including approaches to testing and treatment; avoidance of coercion by government, society or physicians (autonomy).
- Respect for human diversity and for those whose views are in the minority (autonomy, non-maleficence).
- Respect for people's basic intelligence, regardless of their knowledge (autonomy).
- Education about genetics for the public, medical and other health professionals, teachers, clergy and other persons who are sources of religious information (beneficence).
- Close cooperation with patient and parent organizations (autonomy).
- Preventing of unfair discrimination or favouritism in employment, insurance or schooling based on genetic information (non-maleficence).
- Teamwork with other professionals through a network of referrals. When possible, help individuals and families to become informed members of the team (beneficence, autonomy).
- Use of non-discriminatory language that respects individuals as persons (autonomy).

Non-discriminatory language should be used in persons with genetic conditions. Thus, for example, someone with Down's syndrome is best described as a person (or child) with Down's syndrome" rather than a "Down's syndrome child" or "Down syndrome case." Words that dehumanize persons with disabilities or stigmatize them should be avoided.

- Timely provision of indicated services of follow-up treatment (non-maleficence, beneficence).
- Refraining from providing tests or procedures not medically indicated (non-maleficence).
- Providing ongoing quality control of services, including laboratory procedures (non-maleficence).
- If the relative subsequently contacts the registry, however, he or she should be asked to consent to the information being retained and if consent is not forthcoming all the data relating to the individual should be deleted.
- Should the implications of the information held on the register change, renewed consent should be sought.

Confidentiality and access

- Proper security and measures should be in place to ensure the confidentiality of the information contained in the genetic register.
- Access to the information recorded in the genetic register should be on a clear "need to know" basis on the understanding that information acquisition is in the interest of the particular patient.

Research

Identifiable information contained in the genetic register should not be used for research purposes without the consent of the individual to whom it relate.

Planning

Anonymous information obtained from the registers may be used for planning and managerial purposes.

Inappropriate use

- Although there is no evidence to suggest that abuse occurs, the possibility of inappropriate use of information in the genetic registers should not be dismissed.
- Non-directive approach (as explained above), except when treatment is available (autonomy, beneficence).
- Children and adolescents to be involved in decision affecting them, whenever possible (autonomy).
- Duty to re-contact if appropriate and desired (non-maleficence, beneficence, autonomy). Re-contact means keeping abreast of new developments and re-contacting individuals or families on a timely basis regarding any new developments relevant to their health or reproduction, unless otherwise instructed by the individual of family.

Disclosure of Information

Full disclosure of test results must be made and this includes ambiguous test results, new and controversial interpretations, and differences among professional colleagues with regards to interpretation and treatment.

5. GENETIC COUNSELING

Non-directive counseling has two major elements. The first is the provision of accurate, full and unbiased information that individuals and families may use in making decisions. The second is an understanding, empathic relationship that offers guidance and helps people to work towards their own decisions.

Ethical Principles Applied to Genetic Counseling

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

- Informing individuals about the wisdom of disclosing their carrier status to a spouse/partner if pregnancy is intended, and the possibility of harmful effects on the marriage from disclosure (non-maleficence).
- Informing people of their moral duties to disclose a genetic status that may affect public safety (non-maleficence).
- Unbiased presentation of information, insofar as this is possible (autonomy).

6. GENETIC SCREENING AND TESTING

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

Genetic testing is the analysis of the status of a particular gene. A genetic test may establish: (a) a specific diagnosis of a genetic condition in symptomatic individual, (b) the certainty that a particular condition will develop in an individual who is asymptomatic at the time of the testing (pre-symptomatic diagnosis), or (c) the presence of a genetic predisposition to develop a particular complex disease such as cancer or cardiovascular disease.

The main objective of genetic screening and testing is to prevent disease or secure early diagnosis and treatment.

Ethical Guidelines for Genetic Screening and Testing

- Genetic Screening and testing should be voluntary, not mandatory, with the exception noted in the last point below (autonomy).
- Genetic screening and testing should be preceded by adequate information about the purpose and possible outcomes of the screen or test and potential choice to be made (autonomy, non-maleficence);
- Anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened (autonomy).
- Results should not be disclosed to employers, insurers, schools or others without the individual's consent, in order to avoid possible discrimination (autonomy, non-maleficence);
- In rare cases where disclosure may be in the best interest of the individual or for public safety, the health provider may work with the individual towards a decision by him or her (beneficence, non-maleficence, justice);
- Test results should be followed by genetic counseling, particularly when they are unfavourable (autonomy, beneficence);
- If treatment or prevention exists or is available, this should be offered without undue delay (beneficence, non – maleficence)
- If it has been decided to offer carrier screening for a particular disorder, preconception screening would be preferable to antenatal screening.
- Carrier screening in schools should not be contemplated until satisfactory solutions have been identified to the potential problems:

confidentiality, the risk of stigmatization among young people at a very sensitive time in their lives, and difficulties of ensuring that adequate support mechanism were in place.

- Services provided direct to the public should be restricted to screening of carrier status for recessive disorders where there is no significant health implications for the individual.
- Screening should not be applicable to those under the age of 18 or adults lacking the capacity to give consent.
- Newborn screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn (beneficence, justice).

7. INFORMED CONSENT AND GENETIC TESTING

Ethical Guidelines concerning Autonomy and Informed Consent

A. Applicable to clinical practice:

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

- The purpose of the test,
- The chance that it will give a correct prediction,
- The implications of the test results for the individual and family,
- The tested person's option and alternatives,

- The test's potential benefits, and risks, including social, emotional and psychological,
- That social risks include discrimination by insurers and employers (even though this may be illegal), and
- That whatever decision individuals and families make, their care will not be jeopardized.

B. Applicable to research and quality control¹:

The element of valid informed consent includes an explanation of:

- The experimental nature and purpose of the study,
- The reason why the individual is invited to participate, and that participation is voluntary,
- The procedure involved,
- The discomforts and risks (if any) of the test to both the individual and the family,
- The uncertainty of the result of the test for prediction and accurate genetic counseling.
- The possible benefits to others and to science,
- The confidentiality of records identifying the tested individual,

1. MMC Publication on Clinical Trials and Biomedical Research, 2006

- Whom to contact for questions about research or in the event of a research injury or adverse event,
- 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.

C. Consent

Competent adult

- All genetic testing should be based on the free and informed consent of the individual concerned.
- In order to give valid consent the individual must be competent to make a decision, the decision must be voluntary, and it must be based on sufficient accurate information. If there is any suggestion that the consent is not valid, testing should not proceed until this has been clarified.

Incompetent adult

- Nobody should consent on behalf of another adult regardless of that person's mental capacity. Genetic testing may be lawfully carried out without consent, however, if deemed to be necessary and is in the best interests of the incapacitated individual.
- Where testing would be in the best interests of the individual but would not fulfill the doctrine of necessity, the legality of the test is likely to depend upon the individual circumstances. In such cases, special legal advice should be sought.

Child

- A person with parental responsibility may lawfully consent on behalf of a young child who lacks the capacity to make personal decision provided the treatment or the test is considered to be in the child's interests.

Young person

- Any young person with sufficient competence, regardless of age, can independently seek medical advice and give valid consent to testing or treatment. As with adults, the level of competence required to give valid consent will depend upon the gravity of the decision and its wider implications.
- Children and young people who have been brought up in a family affected by genetic disorder may have a higher level of knowledge and understanding of the disorder than would normally be expected in people of their age.
- It is unclear whether a competent minor's refusal of testing which is clearly in his best interest would be binding. In such cases legal advice should be sought.

8. PRE-SYMPTOMATIC AND SUSCEPTIBILITY TESTING

Pre-symptomatic testing refers to identification of healthy individuals who may have inherited a gene for a late-onset disease, and if so will develop the disorder if they live long enough (e.g., Huntington disease). Susceptibility testing identifies healthy individuals who may have inherited a genetic predisposition that puts them at increased risk of developing a multifactorial disease, such as heart disease, Alzheimer disease or cancer, but who, even so, may never develop the disease in question.

Pre-symptomatic testing in the absence of the therapeutic option should be available if the following conditions are met:

- a. The information provided by testing will be used to prevent harm to the person tested, or to spouse, family, prospective children or others.
- b. The person is fully informed about the limitations of testing, including possibilities of uninformative results (result which do not provide any useful clinical information), and inability to predict exact age of onset or (sometimes) severity of symptoms.
- c. The person is mentally capable of giving consent (or the legally authorized representative).
- d. Testing is accompanied by an appropriate counseling programme.

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

Ethical Guidelines for Pre-symptomatic and Susceptibility Testing

- Genetic susceptibility testing of persons with a family history of heart disease, cancer or other common disease of genetic origin should be encouraged, provided that the information from the test can be used effectively for prevention or treatment of the disease (beneficence).
- All susceptibility testing should be voluntary, preceded by adequate information and based on informed consent (autonomy).

- Pre-symptomatic testing should be available for adults at risk who want it, even in the absence of treatment, after proper counseling and informed consent (autonomy).
- Testing of children or adolescents should be carried out only if there are potential medical benefits to the child or adolescent (autonomy, beneficence, non-maleficence).
- Employers, insurers, schools, government agencies or other institutional third parties should not be given access to test results as these results should not influence insurance or employment (non-maleficence).

9. DISCLOSURE AND CONFIDENTIALITY

Disclosure and confidentiality issues are some of the most frequent ethical problems appearing in medical genetics. Because of the possibility of harm from disclosure to institutional third parties, utmost care must be taken to protect confidentiality. However, a genetic diagnosis in an individual may indicate genetic risks in his/her relatives. In those circumstances, the genetic service provider should encourage the individual to ask the relatives to seek genetic counseling. If the individual refuses, especially in cases where effective and affordable treatment or preventive measures are available, the counselor may ethically make direct contact with the relatives, bearing in mind that the information provided should concern only their own genetic risks, and not the genetic status or the identity of the relative who refused to inform them. Counselors should also make sure that adequate follow-up takes place.

Ethical Guidelines concerning Disclosure and Confidentiality

- 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 counseled.
- Test results, including normal results should be communicated to the tested person without undue delay.
- Test results not directly relevant to health, such as non-paternity, or the sex of the fetus in the absence of X-linked disorder, may be withheld if this appears necessary to protect a vulnerable party if prescribed by national law.
- The wish of the 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.
- Information that could cause grave emotional, psychological or social harm may be temporarily withheld. Within the general duty of disclosure, the counselor may exercise judgement about when a tested person is ready to receive information.
- If a couple intends to have children, both individuals should be encouraged to share genetic information with his/her partner.
- Where appropriate, as part of their general duty to educate, counselors should inform people that genetic information may be useful to their relatives and may invite individuals to ask the relative to seek genetic counseling.

- The provision of genetic information to relatives about the family so as to learn their own genetic risks should be possible, especially when a serious burden can be avoided.
- Results of carrier test, pre-symptomatic tests, susceptibility test and prenatal tests should be withheld 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 the country.
- Registries (if any) should be secured by the strictest standards of confidentiality.
- Information may be released to a third party with the individual's consent. A copy of the consent should be retained in the medical record.

10. PRENATAL DIAGNOSIS

Prenatal diagnosis of genetic disorders and fetal anomalies has expanded significantly for hundreds of condition through the DNA analysis or foetal cells, and the increased use of ultrasound and maternal serum biochemical screening (amniocentesis). The purpose of prenatal diagnosis is to rule out the presence of a particular medical condition in the foetus at risk. This information is provided to the couple to assist in their decision-making process regarding the available options, such as carrying the pregnancy to term, preparing for a difficult delivery and for special newborn care, or terminating the pregnancy. Genetic counseling is particularly important prior to prenatal diagnosis and, after a result indicating an effected fetus, to secure fully informed choices. Information about the purposes, benefits and limitations of maternal serum biochemical screening must be given when offering the test, including the fact that any abnormal screening result will need confirmatory testing by invasive prenatal diagnosis and may potentially lead to a decision about abortion.

In Malaysia, the Penal Code² allows a registered medical practitioner to terminate the pregnancy of a woman if such medical practitioner is of the opinion, formed in good faith, that the continuance of the pregnancy would involve risk to the life of the pregnant woman, or injury to the mental or physical health of the pregnant woman, greater than if the pregnancy were terminated, or there is substantial risk that if the child were born, the child would suffer from such physical or mental abnormalities as to be seriously handicapped.

Cultures, religions and national laws differ with regard to abortion of an affected fetus after prenatal diagnosis.

2. Penal Code Revised 1997 (Act 574) in Sections 312-315

Ethical Guidelines for Prenatal Diagnosis

- 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).
- 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 effected fetus (autonomy)
- 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 child with a disorder (autonomy)
- Prenatal diagnosis is carried out only to give parents and physicians information about the health of the fetus. The use of prenatal diagnosis for paternity testing, except in cases of rape or incest, or for gender selection, apart from sex-linked disorders and for selection of inherited characteristics (intelligence, height, hair and eye colour) is not acceptable (non-maleficence)
- Prenatal diagnosis solely for relief of maternal anxiety, in the absence of medical indications, should have lower priority in the allocation of resources than prenatal diagnosis with medical indications (justice)
- Counseling should precede prenatal diagnosis (non-maleficence)
- 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).

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

Pre-test counseling makes post-test counseling (for those with and affected fetus) much less difficult because prospective parents are better prepared.

Counseling Points prior to Prenatal Diagnosis

- 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 family life.
- Possibilities for treatment of the disorder(s) after birth and availability of supportive care.
- Description of the likelihood (risk) that the fetus may have the disorder(s). Risks should be expressed in several way (as a percentage, as a proportion, etc.).
- The possibility of unfavourable test results or of fortuitous or unexpected findings.
- Alternatives available for those with an affected foetus, for example, carrying the foetus 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 of the fetus or early treatment after birth.

- The possibility of ambiguous laboratory or ultrasonography results.
- Information that, because most conditions diagnosed in the foetus cannot be treated before birth, knowing about the existence of a condition may not help the foetus.
- Information that the test does not guarantee a healthy baby, because there are many disorders that cannot be identified before birth, or professionals may not know that a family is at risk of a specific disorder (in addition to the disorder that motivated the examination.)
- The medical risks to foetus and mother posed by the testing procedure.
- Non-medical risks, if any (e.g. to parental employment or health care, where applicable).
- Information that non-invasive screens used early in pregnancy, such as maternal serum alpha fetoprotein screening, may be the first step on the road to prenatal diagnosis and a possible decision about abortion.
- Cost of the test and sources of reimbursement for the mother or couple, if applicable.
- Names and address of genetic support groups or organizations for person with genetic disorders that people can contact if they wish.

11. BANKED DNA

Stored DNA in tissue or blood samples may provide useful information for examination of genetic disorders in families or for research. Information from DNA specimens may be of importance for relatives and not only for the person from whom DNA originates. Therefore, access to store DNA by genetically related family members needs to be considered.

Existing stored specimens or samples, such as those in university or hospital department or collection of blood spots, should not be subject to new rules for consent or re-contact that may be established in the future.

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

- Protection of individuals from possible discrimination by employers and insurers, etc.;
- Possible benefits to the individuals from research findings;
- The possibility of multiple uses of the same in different and unforeseen research projects;
- Possible sharing of samples among collaborators, including international collaborators and commercial entities;
- Advantage and disadvantages for individuals and researchers of removing all identifiers (including coded numbers) from a sample.

A blanket informed consent that would allow use of a sample for genetic research in general, including future as yet unspecified projects, appears to be the most efficient and economical approach, avoiding costly re-contact before each new research project. The ethical principles of informed consent should be followed. The consent should specify that family members may request access to a sample to learn their own genetic status but not that of the donor.

While spouses may not have such a right of access, their concern should be considered. All samples should be used with appropriate regard for confidentiality.

Ethical Guidelines for Access to Banked DNA

- A blanket informed consent that would allow use of a sample in future projects is the most efficient approach. The ethical principles for informed consent should be allowed.
- 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.
- 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 treatment. Donors should inform DNA banks of current address for follow-up.
- When all relatives have deceased and all attempts to contact survivors have failed, stored DNA may be destroyed.
- Spouses should not have access to DNA banks without the donor's consent, but may be informed that DNA has been banked. If a couple is considering having children, it is the moral obligation of the party whose DNA has been banked to provide the spouse with any relevant information.

- Except in 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, school, 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.
- Qualified researchers should have access if identifying characteristics are removed.
- Potentially valuable specimens that could be useful to concerned families in the future should be saved and should be available.

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

12. ASSISTED REPRODUCTION AND MEDICAL GENETICS³

Although not directly related to medical genetics, various types of assisted reproduction technologies are often discussed in connection with genetic counseling. Couples who are at risk of having a child with a genetic disorder may choose alternative options. These may include egg or sperm or embryo donation, or surrogacy. Whichever reproductive alternatives are offered they must be consonant not only with the cultural traditions and beliefs of the country, but also with overall respect for the autonomy of individuals and families.

In this context, reproductive cloning (the creation of a fetus whose genome is entirely derived from another individual) has been rejected by many international bodies, including WHO, has aroused fears in many societies, and is not in accord with currently accepted international ethical standards. Therefore, reproductive cloning is also prohibited in our country.

13. GENETIC REGISTERS

Genetic registers come in various forms but most contain information about people affected by a genetic disorder who have sought advice from a genetic center or genetic unit. Some registers also include information about other family members who may be at risk, even though those individuals may be unaware of their at-risk status and do not know that information about them has been recorded in the register.

3. For further details, refer to the MMC Guidelines on Assisted Reproductive Techniques, 2006

Consent

- The rationale to the workings of a genetic register should be explained to the patient prior to seeking consent to the recording of information about them on the register.
- It will usually be in the interest of the family members to be included on a genetic register provided the information is kept confidential since this may allow them to be contacted and informed of their individual at- risk status.

14. INSURANCE

The perceived problem

- It is feared that all individuals with a positive genetic result will be refused insurance policies or charged excessively high premiums, making people reluctant to seek testing which may be of benefit to them in other ways.
- There is concern that if genetic information is used by insurance companies, individuals will be less willing to participate in research projects or to share the results of genetic test with their primary physician.

Existing disorders

Where an individual is already affected by a genetic disorder the situation will be the same as for any other individual with an existing illness and the information must be revealed to the insurance company on the proposal form.

Family History

Virtually all health and life insurance proposal forms request details of family history and, as with existing conditions, an individual's failure to reveal a known family history of a genetic disorder would be considered a fraud and may invalidate the policy.

Existing genetic information

- Information about a positive pre-symptomatic genetic test or a test for predisposition to a particular disorder must usually be disclosed to the insurance company, although for some types of insurance the information is not required.
- If it is not clear from the information available with a request for a medical report, whether genetic results must be disclosed, the patient should be encouraged to check the policy of the company before the report is prepared and submitted.

Requesting for testing

Mandatory genetic testing for insurance purpose is not permitted.

Interpreting genetic information

- If insurance companies wish to make use of genetic information, they must show, unequivocally, that they have the expertise and knowledge base to interpret the results correctly. Clinical geneticists should be consulted if necessary.
- An independent arbiter should be available to mediate in the event of disputes about the relevance of particular genetic information to the cover being sought.

Confidentiality

Particular care needs to be taken to protect the confidentiality of all genetic information.

Informing patients

Anyone considering genetic testing must be informed of the potential use of that information by insurance companies. It is therefore essential that those providing information about genetic testing are aware of the complexities.

15. EMPLOYMENT

Occupational hazards

- Health professionals asked to perform or to participate in genetic screening of employees regarding occupational hazard, at the request of the employer, should satisfy themselves that the following criteria are met:
- There is strong evidence of clear connection between working environment and the development of the condition for which the screening is conducted.
- The condition in question is one in which an affected employee is likely to present a serious danger to third parties or which may seriously endanger the health of the employees.
- The condition is one the dangers of which cannot be eliminated or significantly reduced by the measures taken by the employer to modify or respond to the environmental risks.

General health information

The benefits to employers of pre-employment genetic screening as a predictor of future health and illness are unlikely to justify the expense at present.

Existing genetic information

- Employer should be encouraged to request, in pre-employment medical reports, only information which is directly relevant to the individual's current fitness for the particular work concerned.
- Where specific information is required on an applicant, but the employee does not want this disclosed to the employer, the health professionals may not selectively omit information from the report without making it clear to the employer that there is additional information which the applicant has asked to be withheld.
- Information about positive genetic test should be expanded to give details of the extent to which the result would affect the individual's ability to perform the tasks associated with his or her work.
- Occupational health physicians receiving pre-employment medical reports showing a positive genetic test should seek specialist advice about the implication of the result before commenting on the individual's application.

Legal protection

Those who are genetically predisposed to disability should be given the same level of protection against discrimination as those with existing disability.

Informing the patient

In order to give a valid consent to genetic testing, the individual must be aware of the possible implication for future employment prospects. It is therefore important that those providing information, including family physicians, are able to advise on the wider implications of genetic testing.

16. PATERNITY TESTING

Consent and refusal

- With the consent of the mother, the putative father and, depending on maturity, there is nothing legally to prevent paternity testing being undertaken on request.
- A blood sample may not be taken, or used for paternity testing, without the appropriate consent.
- Although the legal validity of the refusal of a competent minor is unclear, it will usually be in the minor's interests to proceed even if he or she has given an informed refusal.
- A court may issue a directive for paternity testing to be carried out but this does not authorize the taking of blood without consent. Adverse inferences can be drawn by the court, from a man's refusal to provide a sample for testing.
- The same technique may be used to establish maternity, for example where an individual was adopted in childhood and wishes to identify his or her genetic mother.

- Ethically, health professionals should agree to provide assistance with paternity testing only where this is considered to be in the best of interest of the child.
- Before testing, discussion should take place about the reasons for the request, the implications of receiving the information, and the effect of the information on the child.

17. IMMIGRATION

DNA profiling may be used as an evidence of a claimed family relationship to support an immigration application.

Standard rules should apply to cases in which testing will be sought. This should not be based on the ethnic background of the applicant.

Blood should not be taken or tested without consent. In order to give a valid consent to the taking and testing of a blood sample, information must be provided in a way and in the language that the applicant can understand.

18. INVESTIGATION OF SERIOUS CRIME

Whilst the use of DNA profiling provides an important tool in policing, its value in the detection of crime should not be overestimated.

Those suspected or convicted of a serious criminal offence are required to provide a sample of tissue for DNA analysis, the results of which are maintained on a DNA database. The analyses of those who are not convicted are deleted from the record but of those convicted are retained for future reference.

19. GENE THERAPY AND CLONING

Issues likely to present health professionals with ethical dilemmas are gene therapy and cloning.

REFERENCE

1. Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services, Report of a WHO on Ethical Issues in Medical Genetics, updated edition: 4th October, 2000
2. Human Genetics – Choice and Responsibility, British Medical Association, 1998
3. Penal Code Revised 1997 (Act 574) “Causing Miscarriage; Injuries to Unborn Children, Exposure of Infants; and Concealment of Births.” Sections 312-315
4. MMC Guideline on Assisted Reproductive Technology 2005
5. MMC Code of Professional Conduct 1987 Section 2.1.5 “Induced non-therapeutic abortion”

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