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ATTITUDE AND ALGORITHM FOR PROTECTION AGAINST GENETIC DISCRIMINATION

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SUMMARY:

Genetic information may come from many sources. These sources include a person's family medical history, a clinical examination that diagnoses a genetic disorder, or a scientific test. Genetic testing is sought in various contexts for different purposes. The uses of genetic testing are likely to expand over time as the testing processes become easier to undertake and their practical uses become clearer

Medical practitioners use genetic testing to diagnose patients for treatment as well as for predictive, presymptomatic, screening and prenatal purposes. Practitioners request the various types of genetic tests through request pathways, which may involve referral of the patient to a clinical geneticist and also pre-test and post-test counselling.

The global efficiency of a genetic test is measured by three principal indicators: analytic validity, clinical validity, and clinical usefulness. The analytic validity of a test is determined by its capacity to identify a given genotype with precision and validity. Clinical validity corresponds to the ability of a genetic test to detect or predict the presence or absence of a phenotype or a disease. Clinical usefulness concerns the contribution to clinical decision-making and improvements in individual health that a genetic test can make.

Discrimination based on genetic information is especially pernicious because genetic markers nearly always only indicate an increased chance, but no certainty, that a manifested condition will develop.

People who fear they will lose their job or health insurance because of their genetic makeup avoid getting tested, never realizing the potential benefits of early detection and prevention. They also shy away from participating in medical research, hindering scientific progress and the ability to fully enter this new era of medical promise.

As a result, individuals and our society as a whole cannot enjoy the full benefits and savings that could be reaped from genetic science. Public concern about genetic discrimination will only increase as genetic science advances and becomes a greater part of our medical care.

The risk of genetic discrimination takes a toll on individual health. The most critical benefit genetic testing offers is its potential to improve personal health by enabling individuals to better predict risk and possibly to prevent or delay the onset of serious health conditions.

Key Words: Genetic counseling, Genetic disorder, Genetic information, Genetic testing, Informed consent.

The term "genetic testing" refers to any test that is carried out to obtain information about certain aspects of a person's genetic status by revealing an existing or potential medical problem. Genetic tests are performed at the request of the patient or their legal representative, health professionals or researchers.

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Medical researchers use genetic testing to advance medical or scientific knowledge about how genes influence the health of individuals and populations. Genetic testing for research purposes may be conducted in concert with medical practitioners, who liaise with participating patients.

Individuals generally cannot obtain direct access to clinical genetic testing by laboratories. Thus diagnostic, predictive, presymptomatic, genetic carrier, screening, pre-implantation and prenatal genetic testing must generally be sought through a medical practitioner.

Lawyers and litigants use genetic testing as evidence in criminal and civil cases. In criminal cases, genetic testing may be used to prosecute offenders, obtain acquittals, and to press for reversal of convictions on appeal. Litigants also use genetic testing in civil cases, for example, to establish parentage in family law or succession matters. In the future, genetic testing may also be used by litigants in negligence actions to establish or defend a claim.

Employers may seek to use genetic testing to screen

or monitor employees or job applicants. Although this type of testing is not common in Australia at present, overseas experience suggests that these uses are likely to expand in the future. Employers may seek to conduct genetic testing to reduce workers compensation claims, comply with occupational health and safety obligations, or increase productivity by screening out employees who are most likely to be absent from work due to illness. The testing may take the form of predictive or presymptomatic testing to identify whether an individual who is currently asymptomatic has a gene that increases the likelihood that he or she will develop a disorder as a result of the workplace environment. Testing may also screen for genes or disorders that are unrelated to the workplace but which may render an individual undesirable to an employer.

Insurers may use the results of genetic testing as a component of the underwriting process in applications for personal insurance, where health information is collected to assess the risk that applicants bring to the insurance pool. The testing may take the form of diagnostic, predictive or presymptomatic testing, particularly in relation to life insurance.

Government agencies may use genetic testing to establish kinship or identification.

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A number of normative international documents address genetic testing. Certain documents exclusively explore this subject, whereas others take a more general approach and include specific dispositions on genetic tests.

For genetic testing carried out in a clinical context, the Organisation for Economic Co-operation and Development (OECD) states that informed consent is a prerequisite, that genetic counseling should be available both before and after testing, and that the protection of confidentiality and security of personal genetic information should be ensured.

The 25 Recommendations on the ethical, legal, and social implications of genetic testing, presented by the European Commission, explores the use of genetic testing for both clinical and research purposes. It addresses data protection (confidentiality, privacy, autonomy), protection against discrimination, consent, and genetic counseling.

The World Health Organization in its report entitled Medical Genetic Services in Developing Countries, mentions that participation in genetic testing must be voluntary, informed, and supported by a genetic counsellor.

The World Medical Association Statement on Genetics and Medicine sets out prerequisite conditions for genetic testing. Notably, it specifies that predisposition testing will only be carried out with consenting adults, except in cases where there is existing treatment for the disease in question and where test results would facilitate the quick establishment of this treatment.

Achieving justice in this complex area is not susceptible to a simple vindication of individual rights. Careful consideration of the legal and policy issues thrown up by the use of genetic samples and information requires a wide range of interests to be balanced. Achieving the proper balance is difficult in practice, since various interests will compete and clash across the spectrum of activity.

A balance must be struck in a number of other areas in such a way as to recognize and accommodate broad social interests rather than individual ones—such as in the compulsory acquisition of DNA samples by law enforcement authorities; the ability of researchers to gain a waiver of individual consent requirements; the imposition of restrictions on employers from requiring genetic testing and information from their employees; or in limiting the ability of a person to initiate parentage testing without the knowledge and consent of the child and the other parent.

Scientists have already identified genetic markers for various diseases and health conditions, including cancer, diabetes, Alzheimer's disease, Huntington's disease, cystic fibrosis, and potentially thousands of others. Genetic tests are currently available to identify predispositions to specific conditions, and more are expected as science advances.

Although none of these tests predict with full certainty that a condition will develop, they provide a new opportunity for individuals to know more about the potential risk of disease for themselves and their families.

Once informed about their genetic status, individuals can take proactive steps to protect their health, enhance their well-being, and lower health care costs for themselves and society as a whole.

Discrimination based on genetic information is especially pernicious because genetic markers nearly always only indicate an increased chance, but no certainty, that a manifested condition will develop.

People who fear they will lose their job or health insurance because of their genetic makeup avoid getting tested, never realizing the potential benefits of early detection and prevention. They also shy away from participating in medical research, hindering scientific progress and the ability to fully enter this new era of medical promise.

As a result, individuals and our society as a whole cannot enjoy the full benefits and savings that could be reaped from genetic science. Public concern about genetic discrimination will only increase as genetic science advances and becomes a greater part of our medical care.

The risk of genetic discrimination takes a toll on individual health. The most critical benefit genetic testing offers is its potential to improve personal health by enabling individuals to better predict risk and possibly to prevent or delay the onset of serious health conditions. A predictive test for diabetes risk, for example, could influence an individual to make dietary and lifestyle changes that might significantly improve his health prognosis, lessening, delaying or even preventing the onset of disease altogether.

As more tests are developed, increasing numbers of people will be able to reap dramatic health benefits from genetic tests. Conversely, foregoing genetic tests due to a fear of discrimination means a loss of opportunity to improve one's health.

Individuals' fear of discrimination often leads them to shield their genetic information, even from their health care providers, but this can actually pose new health risks. When people refuse to be tested, or are tested using an alias or other device which keeps the results out of their medical records, they lose the benefit of more complete medical histories which could have enabled their health providers to better diagnose, treat, or prevent the onset of illness.

Doctors are not fully informed, which can hinder patients' ability to get the best care possible and ultimately jeopardize their health.

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Approach to genetic diagnosis.

Genetic diagnosis should be performed base on informed, carefully considered and freely chosen personal decision. To the person undergoing genetic testing should be provided accurate information regarding the nature of the study and the purpose of the test. Under no circumstances should an individual be coerced into testing.

Information provided.

To the person being tested should be provided **comprehensive information** regarding:

- type of disorder or condition for which testing is being considered ,
- inheritance pattern,
- risks of developing the genetic condition in future generations,
- methods for prevention.

Ways of obtaining the information:

- personal genetic consultation
- no direct influence of the genetic professional on the decision making process.
- professional written opinion and conclusion, based on provided genetic consultation.

Consent to obtain a specimen for genetic testing.

The patient should declare that he/she has been informed about the following facts and circumstances:

- it is necessary to provide own biological sample;
- if necessary, biological sample may be obtained by members of his/her family;
- benefits and risks of the particular genetic testing;
- understanding that some genetic testing can involve possible medical, psychological or insurance issued for the patient or members of his/her family;
- the biological sample will be examined, for the purpose of genetic mutation identification.

Indications for genetic consultation.

- A personal or family history of a genetic condition, birth defect, chromosomal disorder, or hereditary cancer;
- Two or more pregnancy losses (miscarriages), a stillbirth, or a baby who died;
- A child with a known inherited disorder, a birth defect, mental retardation, or developmental delay;
- A woman who is pregnant or plans to become pregnant at or after age 35;
- Abnormal test results that suggest a genetic or chromosomal condition;
- An increased risk of developing or passing on a particular genetic disorder on the basis of a person's ethnic background;
- An increased risk of developing or passing on a particular genetic disorder on the basis of a person's ethnic background;
- People related by blood, who plan to have children together.

Regulating the process of collecting genetic material.

DNA material should be collected in relation to genetic disorder prevention, improvement the process of diagnosis. Genetic testing can provide information about person's genes and chromosomes. Most of the times, testing is used to find changes that are associated with inherited disorders.

Genetic procedures should be performed when it is necessary, with regard the benefits of genetic testing. Along with the beneficial element of the testing, it is necessary to be guaranteed the protection of person's human rights.

When a genetic testing is done in a research setting, it is necessary to obtain permission by Research Ethic Committee.

Processing and Analysis of genetic material.

Researchers should consider the social and cultural significance of their research, especially in the areas of complex socially significant characteristics and the genetic characteristics of collectivities. When such characteristics are the subject of research, Research ethics committees should satisfy themselves that there are no contestable or dubious ethical values subsumed within the research protocol.

Researchers must ensure the confidentiality and privacy of stored genetic information or research results relating to identified or potentially identifiable participants. Researchers must keep information provided by participants about family members confidential. Such confidential information must not be revealed either to family members or to persons who are not family members.

The research protocol must specify whether genetic information or genetic material, and any information derived from studying the genetic material, will be stored in identified, potentially identifiable (coded) or de-identified (not identifiable, anonymous) form.

Detailed regulation on results of genetic testing.

The legal framework should regulate the following sets of questions:

- providers of genetic material;
- methods of testing research;
- time period;
- storage of genetic information;
- organization conducting the research;
- information regarding the research team directly involved in genetic material processing;
- ways of obtaining a consent;
- information on the research;
- form of the consent;
- principles regarding revealing of genetic information;
- storage of genetic material;
- genetic samples anonymity.

Access to genetic results and genetic information.

Researchers should consider carefully the consequences of storing information and material in deidentified form for the proposed research, for future research and for communication of research results to participants. Identifying genetic information must not be released to others, including family members, without the written consent of the individual to whom the information relates, or a person or institution which may legally provide consent for that person.

Additional matters needing more detailed regulation include:

- rights and obligations of medical personnel, directly involved in genetic material processing;
- rights and obligations of the person providing genetic sample;
- rights and obligations of person's relatives and family members;
- rights and obligations of other family members and persons who are not family members.

Assessment of genetic risk for patient's relatives. In every population, there is a risk of giving birth to a child with abnormalities.

Some genetic conditions are caused by mutations in a single gene. These conditions are usually inherited in one of several straightforward patterns, depending on the gene involved.

- Risk of **monogenic diseases** is constant and is determined by the type of inheritance as well as the carrier status of both parents.
- Risk of **polygenic diseases** is variable, as polygenic disease is under the influence of multiple genes and there is slight familial inheritance pattern.
- **Determining factors** in the assessment of genetic risks includes the severity of clinical condition, emotional and physical history in the family, birth of child with inherited genetic condition.

Restriction the access of certain categories to results of genetic testing.

- medical personnel, having no direct relation to patient's diagnosis and treatment;
- organizations representatives – patient's employer representatives;
- insurance companies representatives, obtaining information for the purpose of insurance activities;
- representatives of other institutions, except cases when information is obtained for legal purposes.
- identifying genetic information must not be released to others, including family members, without the written consent of the individual to whom the information relates, or a person or institution which may legally provide consent for that person.

Regulation on specific issues regarding information on genetic testing.

- until the stage, when the person providing genetic sample, can freely express personal decision, in all matters regarding the genetic testing;
- after the stage when the person providing genetic sample can no longer freely express personal decision;
- in case of death of the person providing genetic sample.

Limiting the transmission of genetic conditions and diseases.

- **by planning** and performing activities related to prevention of genetic conditions, in particular prenatal and preimplantation genetic diagnosis;
- **by mass genetic screening and selective screening** for detecting treatable genetic conditions;

Genetic disease therapy.

- **conventional therapy** via: limiting risk factors, health diet, replacement therapy, surgical intervention, transplantation;
- **gene therapy** of monogenic diseases, disease antis tress therapy, related to somatic mutations, tumor suppressor gene therapy of cancer diseases, gene therapy of cancer diseases immune system stimulation.

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