

5. LEGAL ISSUES in GENETIC TESTING

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Medical science is advancing rapidly in the field of genetics. Scientists are at the threshold of developing treatments where individual genes may be altered to the benefit or the detriment of the individual. Cloning will allow for a genetic identical twin to be produced.

Information from genetic testing can affect the lives of individuals and of their families. Genetic testing is a complex process and individuals may wish to be tested if:

1. There is a family history of one specific disease
2. They show symptoms of a genetic disorder
3. They are concerned about passing on a genetic problem to their children.

Also, genetic profiles, or “DNA fingerprints” are compiled from the results of DNA testing to identify unique characteristics of an individual. No two individuals (save identical twins) are alike. This information has significant application in the forensic field and in cases involving paternity, and in the identification of victims of disasters and wars.

Moreover, the issue of genetic susceptibility to disease may have implications for employment and insurance. On a fundamental rights level, prenatal diagnosis and screening, if abused, can pose a serious threat to the right to life at inception.

Critical issues, which require legal regulation, include:

- **Privacy** - the rights of the individuals to maintain privacy.
- **Informed consent** - obtaining permission to carry out genetic testing. One must have knowledge of the risks, benefits, effectiveness and alternatives to testing in order to better understand the implications of genetic testing and exercise a choice.
- **Confidentiality** - this concerns the recognition that genetic information is sensitive and should be restricted to those authorised to receive it. Future access to a person's genetic information should also be limited.

The Convention on Bioethics

Existing national laws may regulate these issues relatively to conventional medicine. However, the implications of the new technologies not only on the individual but also on the human species necessitate specific rules.

The Convention on Bioethics and Medicine, 1997, adopted by the Council of Europe, has provisions affecting gene therapy, biotechnological research and cloning. This Convention makes it clear as a basic principle, that the individual is entitled to protection against unlawful interference with the human body, and prohibits the use of all or part of the body for financial gain.

In terms of *Article 1*, States Parties to the Convention are obliged to *protect the dignity and identity of all human beings and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the application of biology and medicine*. In enunciating this principle, the Convention is entirely in consonance with previously existing Human rights treaties. The Convention covers all medical and biological applications

concerning human beings, including preventive, diagnostic, therapeutic and research applications.

The Convention does, however, also address its concern for the protection, not only of the individual, but also of present and future generations. The individual is thus placed in a social context as constituting part of society and of the human race. Nevertheless, the interests are not equal but are graded to reflect the priority attached to the interests of the individual as opposed to those of science and society alone. With reference to the benefits of biology and medicine to future generations, the Convention makes provisions for the necessary legal guarantees to protect the identity of the human being.

The primacy of the human being is expressed in article 2 of the Convention⁷. This is subject to certain restrictions, which largely echo *Article 8(2)* of the European Convention on Human Rights. These restrictions are such as are prescribed by law and are *necessary in a democratic society in the interest of public safety, for the prevention of disorder or crime, for the protection of public health or for the protection of the rights and freedoms of others*.

Thus the restriction based on the prevention of disorder would make it possible for the respect of privacy to be restricted by permitting a judicial authority to order a test to be carried out to identify the perpetrator of a crime.

Protection of the rights of others may, for example, justify an order by a judicial authority for a test to be carried out to establish parentage.

The Right To Privacy.

The right to privacy is a fundamental human right enshrined in international human rights Treaties. Each individual shall

be protected from the unlawful invasion by the State of this basic right and from any State act or authority which would undermine his dignity as a human being.

In consonance with the principle of primacy of the individual and the need to protect him from the improper use of scientific developments, the Bioethics Convention provides protection against the unlawful interference with the human body, and prohibits the use of all or part of the body for financial gain. It furthermore restricts the use of genetic testing.

The Convention in *Article 5* provides quite clearly that no intervention may be carried out in the health field without the free and informed consent of the person undergoing it.

Interventions in the field of research or application aimed at modifying the human genome are allowed on two conditions:

- a) That the intervention must be undertaken for preventive, therapeutic or diagnostic purposes. Consequently interventions aimed at modifying genetic characteristics not related to disease are prohibited.
- b) The aim of the intervention must not be to interfere with the human reproductive cells of a person who has already been born or of that of an unborn child. However it does not rule out interventions which may have unforeseen side effects on the human reproductive cells.

These restrictions are justified in view of the problems related to predictive testing as shall be illustrated further. Predictive testing here is strictly limited to its applicability to the health purposes of the individual. Commercial interests such as those of employers or insurance companies are excluded. Thus genetic testing as part of pre-employment medical examinations are excluded whenever they do not serve a health purpose. However, national law may allow such testing

for the reasons already stated justifying a limitation on the right to privacy of the individual.

With reference to tests which are predictive of genetic diseases, these tests could cover both the detection of the presence of genetic factors for a disease, or a predisposition to genetic disease. Sometimes the predisposition is certain to lead to a disease developing, and sometimes it can only indicate a possibility of the development of disease. In this latter case, early detection would allow for preventive measures such as adapting one's lifestyle or environmental conditions. This process may have advantages, therefore, for the future health of the individual as it would be expected to positively influence one's health. Tests that are predictive of genetic disease would also allow for informed decisions concerning one's offspring.

In this field, the right to know, as well as the right not to know are of particular importance. A complicating factor is that testing generates information not only on the individual concerned, but also on future offspring and on the biologically related family members. The right of privacy therefore involves more than one individual.

An example that can be given is in relation to the Tay-Sachs gene. If two persons carrying this gene marry, then statistically 25% of their children would receive two abnormal Tay-sachs genes which would produce a person afflicted with the disease (as opposed to being a carrier). The privacy of the individual leaves testing and decisions to the ambit of individual choice.

However, it is important to note that the Bioethics Convention prohibits predictive testing for reasons other than health or health-related even with the consent of the person concerned. Consequently, predictive testing in the field of employment or private insurance, for example, which does not have a health

purpose, would imply an infringement of the rights of the individual to privacy. An exception to this could justifiably arise from a work environment which may have deleterious consequences on the individual's health if he/she has a certain genetic predisposition. However, testing would be justified only if there are no reasonable possibilities of improving on working conditions and provided the tests clearly serve the health condition of the individual.

Informed Consent.

I have stated that the right to know as well as the right not to know is of particular importance in this field. Such problems can usually be addressed within the context of the patient-doctor relationship. In particular the patient's right not to know is discussed within the context of predictive testing for serious late-onset diseases for which at present, no treatment is available.

One could argue that what is of little therapeutic value is of no value to the patient either. Yet this paternalistic approach runs counter to recent advocacy of the patient's right to be informed of his/her medical condition.

Of course, there is no right to genetic testing per se. An individual has a right to health care but this would not necessarily imply a right to every diagnostic test not reasonably required for proper care.

It is true, however, that genetic tests are not the only source of information about a patient's condition and standard family medical histories can also shed light on an individual's susceptibility to disease.

Those who favour medical paternalism fear the effects of so-called *toxic knowledge*. For some people, the burden of the

discovery that they are at risk of suffering life-threatening diseases may so depress them that the quality and purpose of their lives would evaporate. However, it is also true to say that this reaction would vary from individual to individual.

There is an alternative to this attitude. The physician can ask patients before testing for one condition, whether they wish to have the information about another condition that will become available from the test. This places the decision within the ambit of the patient's control.

Confidentiality

The issue of the right to know is closely linked with that of confidentiality. Concerns about discrimination in employment or loss of insurance coverage are usually cited among persons refusing to take genetic tests.

Article 17 of the Bioethics Convention as we have seen, only allows genetic testing for health care purposes. The use of genetic testing outside health care, for example, pre-employment medicals, does not fall within this parameter. It is therefore important to distinguish between health-care purposes for the benefit of the individual on the one hand, and third parties' interests, which may be commercial, on the other hand.

As we have also seen, the consent of the individual would not make such tests permissible. Consequently, it would seem that an insurance company is not entitled to subject the conclusion or modification of an insurance policy to the holding of a predictive genetic test. Nor will the company be able to refuse issuing a policy on the basis that the individual applicant has not submitted to a test. Within this context, the insistence of the insurance company would imply a disproportionate infringement on the right of the individual to privacy.

Two cases exist which can be associated to this issue:

In *Katskee v Blue Cross/Blue Shield of Nebraska* the patient was found to have a 50% likelihood of developing breast or ovarian cancer because of her genetic make-up. She had surgery performed to prevent the disease and the insurance carrier denied payment because no cancer was currently present. The Court found that the insurer was responsible for the costs. The decision was based on the probability that the defective gene would cause a problem and in this sense was considered to be an illness.

In another case an insured was denied coverage for medical bills associated with retinal detachment. The insurer based the decision on the fact that the medical problems leading to the detachment constituted a pre-existing condition. In this case the carrier was found responsible because the condition was unknown to all parties at the time the policy was entered into.

It is this concern that knowing of one's susceptibility as a result of predictive testing would automatically void medical insurance policies that is often cited as a basis for refusing to submit to testing. Every genetic abnormality constitutes a pre-existing condition. From an insurer's point of view, a potential insured who tests positive for a particular condition is being insured at a rate not representative of the risk that person holds. Again, standard medical tests and family history generally places insurers in a position to make well-informed decisions about a potential insured person's suitability for coverage.

A case study may illustrate the pitfalls for the individual. An individual, let's call him Frank, a 35-year-old truck driver, fell and hurt his arm and was taken to a local hospital for treatment. He signed routine forms to conduct tests and treatment. As the hospital was also affiliated to the University, the forms

provided for consent for the medical information to be used in ongoing research. Consequently, blood tests included a DNA test. Frank's employer informed the company's insurer of the accident and the latter requested copies of Frank's medical results relating to the accident.

On his release, Frank instructed the hospital clerk to forward all documentation to the insurer. Unknown to him, the genetic screening showed that he was at significantly high risk of developing heart disease.

The upshot of this was that the insurance company, on receiving his medical records, decided that he was too high a risk for the company to continue to insure, thus placing the employer in a position of being unable to provide group coverage. Frank ultimately lost his job.

An interesting sideline to this study was that Frank had also applied for a loan to buy a new house and willingly supplied his medical records to the loan officer. The loan was refused.

In this study, the hospital records did not have a special system to separate the results of the genetic tests from the other medical results. In a sense there was no breach of confidentiality because the patient himself had authorised the transmission of the records to the insurance company. Yet was Frank fully informed of the tests to be carried out on him? Was his consent to testing sufficient to be deemed to cover also genetic testing? Would the hospital be responsible in this case?

Although this case is cited with respect of assessing insurance issues, it does raise difficulties attendant on the matter of informed consent and on confidentiality. An insurer would require full disclosure of any medical knowledge, which would affect the policy at the time of application. Consequently, if genetic testing has been done, the potential insured will have

to disclose the information. Otherwise the policy is void. Thus whilst no company can require genetic testing in order to insure, the applicant is required to disclose a result of a test already performed.

Yet should this matter be left to individual contracting parties? In the Netherlands, for example, federal legislation disallows insurers from requesting or using genetic information for life insurance policies which do not exceed a stipulated value. A number of states in the USA whilst not prohibiting the use of DNA data for underwriting purposes, strictly limit it. By New Jersey statute, for example, health insurers other than life and disability insurers are banned from using the information at all.

In a 1992 report on *Genetic Testing and privacy* the Privacy Commissioner of Canada asserted that Canadians should have “a reasonable expectation of genetic privacy”. Access to such private information as a person’s genetic make-up makes many uncomfortable, and the use of such data can have far-reaching effects.

The disastrous effects of indiscriminate release of information on the individual’s life have been illustrated above. The Bioethics Convention strictly prohibits the communication of test results outside the health field save for the reasons stated in the proviso to article 2 (e.g. for the prevention of disorder or crimes etc). This rationale of this is obvious. It would be more harmful for the individual to refuse to submit to a test about his health for fear of the consequences.

One of the effects that the release of genetic information may cause is discrimination against individuals with less than ideal genetic make up. Certain States have already legislated to preclude discrimination on this basis. In 1992, for example, a New Jersey statute was amended to include “familial status” as a basis for protection from discrimination at the place of work.

The Universal Declaration on the Human Genome and Human Rights

In 1997, the UN approved the Universal Declaration on the Human Genome and Human Rights. Article 6 of the Convention clearly states that “No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.”

Discrimination on this basis is unlawful and violates the basic protection and freedoms to which an individual is entitled.

The **Maltese Constitution** in *Article 45* prohibits discrimination on the basis of race, place of origin, political opinions, colour, creed or sex. This definition would not include discrimination based on “genetic characteristics”.

These are some of the legal problems encountered in the field of genetic testing. Problems do exist and call for immediate regulation. As in other areas, the law is seriously lacking. Issues of privacy, confidentiality, information, sanctions and compensation cannot be left to analogy but must be specifically addressed.