# 3.
# Confidentiality, Privacy and Genetic Testing

**Alfred Cuschieri**

## Introduction

About 25 years ago, when I first began practising clinical genetics in Malta, I wondered why I rarely saw cases of Huntington's disease. It was not because they did not exist or were rare, but because they were not referred for genetic counselling. Psychiatric consultants counselled me that if I wanted to see cases of Huntington's disease, I should visit Mt. Carmel Hospital. At that time Huntington's disease was considered to be a terrible affliction that happened to strike a particular individual, and often the family conveniently overlooked the occurrence of similarly affected relatives. The existing risks were often hidden from children, even when they grew up to be adults and themselves passed on the disease. It was not right to discuss such matters within the family, and much less outside it. This was an example of strict observation of the individual's and the family's privacy. The presence of Huntington's disease in an individual was highly confidential information and was frequently camouflaged by the presence of pulmonary, cardiac, or malignant diseases, which were often quoted as the causes of death.

This extreme picture of confidentiality and privacy has changed dramatically as people became more knowledgeable about health, and hereditary diseases, about genetic testing, prevention of genetic disease, gene therapy, the human genome project, assisted fertilization and cloning. Genetic testing is now being increasingly employed for diagnostic purposes in a wide variety of conditions ranging from

congenital defects in babies to adult onset neurological disorders. It is used for pre-natal and pre-implantation diagnosis, pre-symptomatic diagnoses in persons at risk, for identification of asymptomatic gene carriers, and for prediction of susceptibility to certain diseases such as breast, lung, and colon cancers cardiovascular accidents and other common diseases. Genetic testing is, in a way, different from other medical laboratory tests because it has profound consequences regarding present and future health and longevity, far-reaching social effects regarding marriage and offspring, and most importantly broad implications relating to whole families rather than to individuals [1]

Many people are willing to have genetic tests to provide information about their health status, although they might not fully understand the profound implications of the test results until these are explained to them. The people who want to know are not only the individuals affected by a disease condition, but also their relatives their sons and daughters, the fiancées of engaged offspring, uncles, aunts and cousins and even totally extraneous persons or bodies, such as insurance companies and employers. Depending, of course, on the circumstances of particular cases such individuals may claim that they have a legitimate right to information that directly or indirectly relates to them. The ethical problems regarding confidentiality and privacy are to decide whom to include within the limits of confidentiality and under what circumstances they to be included. The relatives of an individual who tested positive for a genetic condition not only claim that they have a right to know of any results that could affect them but they themselves become entitled to their own privacy and to the confidentiality of their own test results.

## Confidentiality of patient records

Emphasis is now being placed on the inclusion of every detail of investigation and therapy in a person's medical history. It is unethical not to record the results of clinical examination, of clinical laboratory, radiological or other investigations and of surgical interventions. Medical records should include all information about a patient including genetic results, as these could be important in the care of the patient and of other family members in the future (2). However, it has also been argued that certain genetic test results should be excluded from the patient records by virtue of their delicate nature and their far-reaching implications relating both to the persons involved and to their families. Persons who had certain genetics tests performed on them have also expressed fears about the confidentiality of their results. Do such fears on the confidentiality of sensitive issue being included in the medical records imply that the confidentiality of these records is not in fact being adequately safeguarded? The ethical issue of safeguarding the confidentiality of patient records needs to be carefully assessed. The currently prevailing attitudes of all those involved in safeguarding this confidentiality need to be evaluated and certainly will have to change. There appear to be several serious misconceptions regarding confidentiality of patients' records. Confidentiality means that the information belongs to a particular individual and is available only to authorized person. However, this may not be the prevalent concept of confidentiality. It is enough to look at the cover of patients' medical records of the Health Service in Malta that warns in bold letters "CONFIDENTIAL: NOT TO BE HANDLED BY THE PATIENT". Does this mean that everyone else, except the patient, is entitled to see the records?

Although genetic testing is sometimes considered to raise special ethical issues regarding confidentiality and privacy, these are not really different from other confidentiality issues

in medical practice. The general principle that the patients' confidentiality and privacy are to be respected applies also to the results of genetic testing. The criteria regulating disclosure of the results of genetic testing is not much different from disclosure of other forms of medical information. Genetic information is certainly a new concept arising from a new science but it raises the same old dilemmas regarding disclosure of information, value of the information, and who owns the information (3)

**Ownership of genetic information.**

There can be no doubt that the results of genetic tests belong to the individual tested. Individuals have the right to control the use of all medical information about themselves, including genetic information (4). The individual, or his or her legal guardian in the case of children, have a right to determine to whom that information is passed on. This might seem clear enough but problems and conflicts do arise. In paternity testing the person paying for the tests might think that he or she has ownership of the test results and may therefore think that he or she has the right to determine whether or not to pass the information to the partner. In fact this is not so. Both partners who have consented to being tested have an inalienable right to know the test results. The father is not free to withhold the information from his partner if she has participated in the testing procedure. It is also unethical for a person to perform paternity genetic tests regarding adolescent sons or daughters without their specific consent, although the alleged father may be the legal guardian of the individuals who are officially considered as minors.

**Informed consent**

Doctors may feel quite secure that they are authorised to disclose information if they have the consent of the individuals

tested, particularly if this is in writing. If fact, however, one must be aware that having a written consent might convey a false sense of security. It is entirely dependent on whether the individual consenting to the disclosure of the information fully understands the implications of his or her consent. An individual giving consent to a genetic test might not realise the implications of that test to the rest of the family or its implications in taking a medical insurance. This means that consent should be truly informed, that the doctor has explained that revealing the test result might work against his or her own interests, and might result in discrimination by insurance companies or at work by failure to be find employment or to be given a promotion. The doctor has the responsibility to point out these potential consequences even if the doctor might not be involved in the actual passing on of the information at a later stage.

A signature at the bottom of a statement agreeing to a genetic test does not constitute informed consent. It is merely a measure of protection for the doctor and not for the patient whose interests we are in duty bound to observe, if only by virtue of the ancient principles enclosed in the Hippocratic oath. Consent is merely the confirmation that one agrees to have a particular test or other procedure being done. It does not provide any confirmation that the individual understands its possible consequences or dangers. Very often an individual turns up for genetic testing with a particular purpose, perhaps to obtain definitive confirmation of a clinical diagnosis or to qualify for some particular aid or benefit, but does not usually realise that the test might not provide the conclusive information that was desired. An individual who agrees to have a genetic test is unlikely to anticipate the problems that may arise concerning his or her family, but the doctor who is consulted has the responsibility of anticipating the problems that commonly arise and inform the client accordingly. The information that accompanies the consent is a moral

responsibility of the doctor and is an integral part of ensuring the privacy of an individual.

**Confidentiality in relation to third parties.**

Third parties who may be interested in obtaining information concerning the test results of others may be divided into two broad categories: (a) employers, insurance companies and other agencies; (b) relatives and family members. The reasons for which the two groups require the information is vastly different. In the first it is related mainly to business and profit-making of the third party, while in the second it is a matter of personal health.

Currently, any insurance company or employer has a right to request a genetic test, just as they have a right to request a medical examination. Informed consent is always required. The problem of privacy does not arise provided there has been truly informed consent, and that the test is used solely for the purpose for which the consent was given. The ethical implication of genetic testing for insurance companies and employers is that they may encourage or perpetuate discrimination against individuals, making the issue of confidentiality of genetic information even more important (5,6). It is not the purpose here to question the ethics of insurance agencies in demanding genetic information, on the basis of which an insurance policy may be refused or subjected to a heavier premium. However, I must point out that the danger that discrimination against an individual or even a whole family might sometimes be based on apparent or perceived risks, resulting from unknown significance of a variation from the 'normal' genotype. The relevance and consequences of possible discrimination varies in different countries depending on existing laws and systems and alternative provisions for health care and pensions.

**Confidentiality in relation to other family members.**

In genetic disorders we are confronted with the situation where the discovery of a genetic condition in one individual has health and social implications for other family members. Do the other family members have a right to know of the risks to their health in order to enable them to undertake preventive or therapeutic measures? Here is a situation where an individual's right to privacy and the right of others to know both weigh heavily, and it is not possible to discard one in favour of the other. Fortunately such situations do not commonly arise. As part of the counselling procedure, affected individuals are told of the importance of volunteering the information to their relatives who might be unaware of the risks facing them. In many cases the individuals comply with the recommendation of informing their relatives, who can then seek medical help. However family feuds unfortunately exist, and sometimes one is faced with the situation where an individual does not want even his sons or daughters to be informed or his own brothers or sisters to know of the genetic risks affecting the whole family. The situation here is a very delicate one, which requires careful assessments of the how great is the risk to the health of the relatives, and how urgent it is to take immediate steps. There is no simple answer to these dilemmas and one has to act very discretely according to the circumstances of each case and adopt a carefully selected strategy to inform relatives of their risks while preserving the confidentiality of the individual tested.

Sometimes, however, the conflict of interest between relatives does not stem from animosities. A person who is at 50% risk of being affected with Huntington's disease may not wish to undertake any pre-symptomatic tests and to prefer to let nature take its course and to worry about the condition only if and when it strikes. This attitude is quite understandable. On the other hand, the person's son or daughter, being of

marriageable age, may wish to know decisively their genetic status prior to marriage. A positive result would imply a positive result also for the parent, and would thus constitute a breach of his privacy. Such a situation can often be resolved by careful and sensitive counselling adopting once again a careful strategy for preserving the privacy of both parties. However a hard core of difficult cases may still persist.

A problem of confidentiality and privacy also crops up when the affected individual cannot, for some reason, pass on the information to his or her relatives, but gives his informed consent and authority to the doctor to convey the risks to the relatives. The confidentiality of the tested person has not been breached but imparting the information to relatives who were previously unaware of the condition may be interpreted as a breach to their privacy. This situation acquires even greater relevance when one considers that in some disorders, notably familial mental retardation, the relatives might be pre-mutation carriers, who are still developing a mutation that has not manifested itself as a clinical disorder, and will not manifest itself in the offspring of the individual but will certainly occur in subsequent generations. It is not a foregone conclusion that unsuspecting individuals may want to know that they are at risk, even if remedial or precautionary measures are available. In some cases the presence of a genetic risk may still be interpreted as a family stigma bearing with it undesired social consequences that an individual would rather live without.

The concept of genetic stigmatisation is still consciously or unconsciously present. Although we may declare ourselves strongly against it, the underlying fear of discrimination lingers in the minds of affected persons and prejudice in the minds of others. The changes brought about by genetic tests and by the whole burst of genetic knowledge necessitates a corresponding shift in pubic education. Fears have been expressed that "privacy as we know it is dead", (7) and that

the combination of scientific breakthroughs, commercialisation of the genome, and ways of dealing with medico-social problems will accelerate the use of genetic data with the result that others may come to know more about an individual than that person knows of herself or himself. However, our genome does not destroy our privacy. Confidentiality and privacy are social issues and it is up to us to preserve and develop the existing ethics of responsibility to care and to extend the existing ethics of privacy to keep pace with the increased knowledge of the Human Genome. (8 )

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