Catholic Bioethics

Joint Bio-ethical Committee of Great Britain and Ireland

The Ethics of Genetic Testing

A Paper prepared for the Joint Bio-ethical Committee of Great Britain and Ireland

In the last few years there have spectacular advances in genetics. As more and more is known about the human genome, the impression has been given that man is totally predetermined by his genes and so is a prisoner of his destiny. It is believed that if our book of life could be read we would know the essence of life. This genetic myth is scientifically unacceptable and ethically dangerous.[1] Yet this goal drives much of the research in genetics. The danger is that genetic testing may be no more than a step toward achieving this end as it seeks to reveal genetic defects without having the means to correct these flaws. In addition, predictive tests are uncertain, except in the case of disorders such as Huntington's Chorea, and only show a faint risk of contracting the disease. Thus the motivation behind some of this work is questionable. While it is obvious that it through genetic research cures will be found, genetics like medicine must remain the servant of the human reality. However, it is important to understand that the tests at present carried out and their purpose and results.

1. The Scientific Facts of Genetic Tests

Genetic testing is used to identify variations in the DNA sequence that correlate with a disease or higher risk of developing a disease.[2] This kind of test can be used for diagnosis before any symptoms of disease are recognisable and to determine the personal risk. It is important to understand the difference between mongenic disease and polygenic gentic diseases. Mongenic genetic disease is a disease that is caused by a single gene. On the other hand polygenic genetic disease is a disease that has more than one cause. It may be a combination of defective genes, or that external circumstances are necessary to trigger the disease.

[1] Le mythe du gène, support du programme de la vie, est tel que cela conduit à l'illusion qu'une connaissance parfaite du génome d'un individu donnerait accès à la réalité et au destin de la personne. C'est cette image à laquelle renvoie une métaphore comme celle du grand livre de la vie dont il suffirait de connaître l'alphabet et la syntaxe génétiques pour parvenir à l'essence de l'être. Or, une telle conception est scientifiquement inacceptable et éthiquement dangereuse. Comité consultatif national d'éthique pour les sciences de la vie et de la santé, Avis n° 46, Génétique et médecine : de la prédiction à la prévention, Paris, 30 octobre 1995.

[2] Genetic Testing is testing for variations in germline DNA sequences or for products/effects arising from changes in hheritable sequences, which are predictive of significant health effects. *Towards Quality Assurance and Harmonisation of Genetic Testing Services in the EU*, Joint Research Center EU, 2003:5.

The different tests are:

Diagnostic testing is usually triggered by symptoms and signs that suggest a genetic cause. In this case the test is performed to confirm, refine or exclude a clinical diagnosis.

Predictive testing is used to establish the risk to a person with no symptoms of developing a genetic disorder in the future. There are two forms of predictive testing:

Presymptomatic testing looks for a mutation in a healthy individual that will almost certainly lead to the occurrence of symptoms, i.e. there is a high risk of a genetic disease such as Huntington's chorea.

Predisposition testing looks for gene mutations that provide a probability of the occurrence of the disorder. The problem is that many common diseases e.g. heart disease, diabetes, atrophy, Alzheimer's, are polygenic in the sense that the gene mutation will only take effect if other factors are present.

Carrier testing seeks to find a gene mutation for a recessively inherited or X-linked disorder i.e. carried in the female line that will not affect the person, but could effect the descendents.

Prenatal testing clarifies if the foetus carries certain mutations. This may be carried out because of the age of the mother e.g. for Down's syndrome, or rarely for single gene disorders.

Genetic screening means that predictive testing is offered to the whole population or sub-populations for specific disorders. The only group programme established at present is for the newborn. It could be used to establish the presence for example of sickle cell anaemia in people of African origin.

Preimplantation analysis follows in vitro fertilisation procedures for genetic mutation. It is carried out on one or two cells removed from an early embryo.

Pharmacogenetics seeks to identify DNA variants that affect drug response. Adverse drug responses affect up to 7% of hospital patients and it is thought that such a test could forward of these side effects.

Genetic Tests are also used for:

Disease sub-typing e.g. to identify different types of cancer

Identity testing for forensic and criminal law applications.

2. The Ethics of Genetic Testing

The general secular approach to genetic testing is clear from the paper of the European Commission Expert Group that broadly described Genetic Testing as "any tests that

yields genetic data" and regarded this data as part of health information.[3] From this they conclude that genetic testing:

- should be protected in the same way as other medical data.[4]
- its clinical application must be realistic, freely available and regarded as an integral part of health service provision
- should never be imposed [5]

Crucially, they said for tests to be meaningful "*the condition screened must be serious, the test highly predictive and follow-up actions [genetic counselling] must be available in terms of healthcare interventions (including reproductive choices)*".[6] Therefore genetic tests raise serious ethical questions.

a) Is Genetic Information the Same as any other Medical Data?

While it is certainly true that genetic information is medical data, it is questionable if it is the same as any other medical data due to many genetic tests being predictive rather than indicative in character. Such predictive tests only raise the possibility of disease. When this is combined with the danger of discrimination based on such probabilities, genetic predictive information is not the same as any other medical data. Likewise, those who have a family history of a genetic disorder such congenital heart disease, may be discriminated against and their families suffer as a result. Therefore, genetic information requires special protection.

b) The Predictive Capability of Genetic Tests

It should not be surprising that the predictive capability is crucial when most genetic diseases have a polygenic basis where many gene-environmental interactions are involved. This means that the presence of a certain gene sequence does not always lead to the gene based disease. In addition, the test may produce both false positives and false negatives results. When this is combined with an unacceptable level of error[7], a lack of certified reference material[8] and in some cases a lack of clinical utility[9], the disturbing tendency to overestimate the clinical utility of genetic tests is serious.[10] In addition, the detrimental side effects of the test must also be taken into account.

The secular solution to these problems has been a call for external quality assessment to assure a higher quality of test results and a database of rare diseases, the development of certified reference materials, and to set up a review board to review the clinical utility of

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[3] European Commission Expert Group, *The 25 recommendations on the ethical, legal and social implications of genetic testing*, Brussels, 2004:8

[4] Idem p. 15.

[5] Idem p.12.

[6] European Commission Expert Group, *The 25 recommendations on the ethical, legal and social implications of genetic testing*, Brussels, 2004:13.

[7] *Towards Quality Assurance and Harmonisation of Genetic Testing Services in the EU, Joint Research Center* EU, 2003:15-18.

[8] Idem p.19.

[9] Idem p.26 – 28. The problem here is that it must be established that a particular DNA sequence is relevant to the disease in question..

[10] The EU Joint Research Center and the European Group on Ethics is science and new technologies both note this tendency. Technically, clinical utility is based on the benefits and the risk of a false result. Idem p 27.

tests.[11] These measures are designed to remove as far as possible the risk dimension of these tests.[12] However, some risk will always remain. *The fundamental principle here is that risk must be balanced by appropriate benefits for the person undertaking the risk.* Hence where the predictive capacity is high and the risk severe as in Huntington's chorea such tests are justified. Where the predictive capability is low, even though the risk the disease would present is severe, such tests are of doubtful morality.

c) Genetic Tests Must Never be Imposed

The secular reasons that support non-compulsory genetic testing are based purely on autonomy. This fails to take into account certain other factors. Genetic tests reveal not only information about oneself, but also the family genetic tree. This genetic tree may be damaged in such a way that through a recessive gene a genetic disease is transmitted. To this must be added the difficulty in predicting the gravity of certain conditions such as Down's syndrome. Yet prenatal diagnosis is already connected to campaigns to eradicate such "cursed lineages". This raises serious questions about the purpose of genetic tests, and the pressure that could be brought to bear on certain couples not to reproduce. Even if this is not intended, the purpose of genetic testing to reassure a person that they do not have a genetic disorder must be balanced against the outcome of a positive test. The discovery that in the future there is a likelihood of an untreatable condition is not necessarily beneficial to the person. It is these reasons that compliment the autonomy-based argument that genetic tests must never be imposed.

The Use of Genetic Information

The ethics of genetic tests cannot be judged on risk alone. The use of such results is an integral part of the morality of testing when it is evident that present medical expertise cannot resolve the medical problem exposed. Genetic information may be used to discriminate. Expert groups stress the importance of genetic counselling.[13] The form this counselling takes is non-directive even though the use of genetic counselling for eugenic purposes is already practiced[14]. Likewise gender issues may arise in societies where women and men are given different rights and privileges. Its use in this way is immoral and its extension because of compassion or racial purity is abhorrent.

Studies involving the use of genetic tests in the workplace have already shown the flaws in the use of genetic results[15]. The European Commission Expert Group holds the fundamental principle that genetic information should not be used in ways that disadvantage or discriminate unfairly against individuals, families or groups in either

[11] Idem Conclusions p. 55.
[12] Technically, clinical utility is based on the benefits and the risk of a false result. Idem p 27.
[13] For example "in the context of healthcare, genetic testing be accompanied by the provision of key information and where appropriate, by the offer of individualised counselling and medical advice (in the case of highly predictive genetic tests for serious disorders, the offer of specific counselling should be mandatory and patients should be strongly encouraged to take advantage of it). European Commission Expert Group, *The 25 recommendations on the ethical, legal and social implications of genetic testing,* Brussels, 2004:14.
[14] For example, in the case of Down syndrome genetic counselling promotes abortion.
[15] Ethical Aspects of Genetic Testing in the Workplace, European Group on Ethics in Science and New Technologies, 28th July 2003.

clinical or non-clinical contexts, including employment insurance, access to social integration and opportunities for general well-being.[16] This principle is based on the dignity of each human person. While such a principle resists the modern tendency to submerge each person's uniqueness in a universal model of what is a human person,[17] it does not clearly take into account the rights of others who will be affected by a genetic disease. Thus a person who is seeking life assurance is responsible to reveal serious genetic predictions. However, the danger of discrimination because of a genetic condition is very real, with the sufferers being denied work, insurance or basic rights. The fundamental principle that genetic information should not be used in ways that disadvantage or discriminate unfairly must be balanced by solidarity. This demands that in a just society the weak are supported by the strong. The classical solution on an individual level is distributive justice based on the principle that individual rights always have to take into account the need of my neighbour.[18] The Common Good and the fundamental value of each person must be respected.[19] All must be given the chance to reach their potential. But the vulnerable fear a society that worships personal fulfilment. [20] The eugenic option as a solution to genetic disorders is already promoted and used by some in the medical profession. Such solutions are disastrous for society as their basis is genetic purity.

The Clinical Utility of Genetic Tests.

While it is hoped that in the future gene correction will be possible, at present no successful splicing of human genes to correct a genetic defect has taken place. Efforts have been made to correct the genetic defect that underlies cystic fibrosis without success. This raises the question of undergoing genetic tests without any hope of correcting genetic faults, i.e. are such tests clinical useful? The answer depends on the disorder. In the case of monogenetic diseases a test can be justified on the basis of whether the person wishes to pass such a disease on to their dependents. However, in polygenetic predictive testing expert opinion is such tests are of doubtful clinical usefulness because of their unpredictability. Obviously such tests have a serious research use, but it is the future of that person that is at risk and great care must be exercised to see

[16] European Commission Expert Group, *The 25 recommendations on the ethical, legal and social implications of genetic testing*, Brussels, 2004:14.

[17] Cellule de Reflexion Bioethique, Commentaires sur un avis du Groupe europeen d'ethique relatif aux aspects ethique des tests genetiques dand le cadre du travail, COMECE, Brussels 10th October, 2003.

[18] Solidarity is ordained to the common good which also gives the particular groups in society a just framework within which to work". The ethics of social aspect of life was summarised by the CDF in three principles. First, the human person is the responsible agent of social life. Second it is solidarity that obliges that person to the common good of society at all levels. Third, subsidiarity indicates that society must not substitute itself in areas where the individual can function well

[19] Seule cette notion de solidarité envers les personnes vulnérables permet de parvenir à des règles juridiques équitables et de leur apporter un fondement. C'est la reconnaissance de cette valeur fondamentale qui fait apparaître discriminatoire toute volonté d'écarter de l'emploi des personnes aptes à exercer leur profession mais présentant des caractéristiques génétiques qui peuvent faire craindre qu'elles développent ultérieurement une maladie Les enjeux ethiques et culturels des tests genetiques, Cellule de Reflexion Bioethique, COMECE, Brussels, 27th February,2004:3

[20] Redistribution through taxation is repugnant to many who see it as a punishment of those who work hard and an encouragement for others to do as little as possible. Solidarity: The Interaction between Individuals and Society, Peter Jeffery CSSp 2004:1.

that they are not damaged either now or in the future. Curiosity alone does not justify such tests.

Proposals

The authors propose that the Joint Bioethical Committee adopt the following:

- Genetic testing for clinical applications must be realistic, and should never be imposed
- Genetic tests are moral when the condition screened is serious, the test highly predictive and follow-up actions are available in terms of healthcare interventions.
- The risk of a test must be balanced by appropriate benefits for the person undertaking the risk.
- Predictive testing with its wide margin of error cannot be acted upon. Such tests could be justified if they have a serious research use, but serious precautions have to be taken to avoid harm to the person being tested.
- The use of genetic information involves on one side the duty to disclose to appropriate persons genetic information that might seriously affect them, but on the other side such information should not be used in ways that disadvantage or discriminate unfairly against individuals, families or groups in either clinical or non-clinical contexts, including employment insurance, access to social integration and opportunities for general well-being.
- The eugenic option as a solution to genetic disorders is disastrous for society and morally wrong as its basis is a search for genetic purity.

Bishop Christopher Budd
Fr. Peter Jeffery CSSp

Documents

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