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# Editorial

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### Ethos and ethics in genetic diagnosis

Some 650 diseases can be now be identified through techniques for genetic diagnosis and our perception of disease is changing as a result. Clinical symptoms are no longer essential for a diagnosis. Gene analysis makes it possible to detect disease at all stages of life. Those conditions which are marked by anomalies of genes or chromosomes can be detected not only in adults, but in the fetus and even in single cells obtained from early embryos and blastocysts. Not only can existing diseases be diagnosed, but the appearance of degenerative diseases later in adult life can be known with certainty and for some types of cancer, the probability of developing a tumor can be predicted. As we accumulate data on the human genome and acquire ever-more sophisticated tools to interpret the data, the number of tests available will grow and probably encompass the 5000 or so conditions with a genetic origin. It will be more difficult, but not entirely impossible, to find solutions for the identification of individual patient risks from conditions that are polygenic and multifactorial, with include a number of common chronic pathological processes including hypertension and dibetes.

For the laboratories involved in molecular and genetic diagnosis, this type of testing will profoundly change the relationships between clinician, laboratorian, and patient. Molecular diagnosis changes the relationship of the individual to his disease and to the medical and scientific community dealing with the disease. The individual patient's perception of himself or herself and the relationships with society and social perceptions can be deeply affected by this new capacity for genetic diagnoses. An additional factor will be the payment for the tests, since it is unlikely that all health insurers will be able to reimburse the costs of genetic screening and testing. Today, a large part is financed by charities and state research funds - tomorrow, access may be easier for only the more wealthy patients.

There can be little doubt that for the genetic diagnoses for which there are definite forms of treatment, the situation is ideal. The test procedure simplifies the clinical examination, eliminates clinical doubt, and allows early instigation of the appropriate treatment. A perfect example is hemochromatosis of genetic origin. The excessive absorption of iron results from a defect in regulation of absorption of iron, the symptoms of which develop over years, with a variety of clinical features : chronic asthenia, arthropathy, skin pigmentation, with cirrhosis, diabetes or cardiomyopathy in later stages. The identification of the gene which is altered in hemochromatosis has radically simplified the diagnosis of this condition. For a patient with an increased saturation of transferrin, the diagnosis of hemachromatosis is made simply by demonstrating that the patient is homozygotic for the mutated form of the gene (C282Y + /+ ).

Hemochromatosis is a remarkable illustration of pathological condition whose clinical management has benefited in record time from a fundamental discovery in molecular genetics. It is something of a paradox that the treatment of the disease by bloodletting is almost medieval in approach, yet the diagnosis is definitely a 21 st century approach.

In other conditions, a diagnosis can be made certain by identifying one or more mutations but this approach does not give an indication of the severity of the disease nor of the nature of the symptoms which will appear. The value of the diagnosis is less when there is no specific treatment, or if there is controversy concerning the effectiveness of the treatment or for measures that prevent the progress of the condition.

Concerning serious or incurable illnesses, the localization of a familial mutation opens the possibility for an antenatal diagnosis. This allows the pregnancy to proceed in the knowledge of the likely outcomes for the child and the family. In some cases, the parents may choose for the pregnancy to be terminated. An alternative approach in some countries is to use in vitro fertilization, with the genetic testing performed on single cell biopsies from blastocysts, with implantation of the blastocysts not showing the gene marker concerned. Certainly this reduces the numbers of fetal deaths and late abortions arising from a number of genetic disorders, but the risk of


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eugenic exploitation of the procedure. Also, this approach can create the impression in society that gene-related disorders, many of which result in handicap, are avoidable. This is far from reality, as is should be clear from the "spontaneous" mutation rates of the genes involved.

For the genes that are predictive of the future development of cancers or degenerative diseases, other questions can be raised. Is it useful for a person in apparent good health to know that this state of well-being will probably not continue, but with no clear indication of when or where the change will happen? How does this knowledge affect the individual's personal, family, social and professional functioning?

For some, the confirmation of presence or absence of the disease makes it possible to eliminate the inevitable anguish of uncertainty. It can help to better prepare the future. However, this is not possible unless the genetic diagnosis is made in a multidisciplinary context. The molecular biologist has to work in a team combining geneticists, psychologists, and other specialists in health disciplines and social support. Notions of guilt and responsibility have to be carefully managed in genetic counseling, in order to avoid rejection of carriers or affected individuals in their social or marital contexts.

Genetic tests are certainly fascinating in terms of science. Despite the precision of the diagnosis, it has to be remembered that they are only a diagnostic description - and often of conditions that have no remedy or effective treatment. Nonetheless they do give cause for hope that mechanisms of cause and effect in gene mutations will be better understood and controlled at sometime in the future.

In the way that genetic testing is performed, it is important to maintain the key importance of what is in the best interest of the patients. Testing has to be regulated to ensure that it does have a medical purpose. The medical purpose can differ from the purely scientific interest, in that it is intended for a particular person and only that person. Investigational decisions that deal with the investigator's perceptions of the "greater good" of society or the development of techniques with commercial potential can sometimes interfere with the logic of the decision-making process, however. The investigation and management of the patients with genetic disorders has to be made with reference to clinical research practice and basic humans rights.

This has led laboratory professionals to undertake a process of deliberation in order to identify the key principles of genetic testing that are essential and which should be shared with society. For these reasons IFCC has set up a committee on ethics (Chair, Professor Leslie BURNETT burnett@med.usyd.edu.au) whose aims are to increase awareness among Laboratory Medicine Professionals of ethical issues ; to encourage the practice of Laboratory Medicine to the highest ethical standards ; to develop position papers on appropriate ethics policies issues ; to provide a voice for Laboratory Medicine on ethics policies ; to link Laboratory Medicine, ethics and the public interest.

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