Commercial gene testing: the need for professional and public debate

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Abstract
Commercial gene tests are currently available in the UK and the range of tests offered is likely to increase. While knowing their genetic status may bring benefits to individuals and their families, genetic testing also brings its own unique problems. Commercial ventures may do little to resolve these and adequate safeguards are needed to ensure that clients undergoing testing are not disadvantaged. The availability of gene tests can only serve to heighten public awareness of the relevance of genetics to health care, and there is likely to be an increased demand for information and advice from healthcare professionals before, and following, testing. Nurses should be prepared to acquire adequate knowledge to maintain their role as health educators, and to participate in public and professional debate on the issues that commercial testing highlights.

A small advertisement appeared in the pages of a popular women’s magazine in the summer of 1995 targeted at women who were planning pregnancy (Cosmopolitan, 1995). The advert was offering an ‘inexpensive’ test (£65 per person or £98 per couple) to detect carrier status of a genetic disorder, using a mouthwash sample which could be collected at home and then posted to the testing laboratory. The results, with a full explanation, were promised within days, with the possibility of further advice and information from a clinical genetics consultant offered at no extra cost. The current information pack for prospective clients indicates that face-to-face counselling is only available in London.

The disorder in question was cystic fibrosis. It is the most common autosomal recessively inherited genetic disease in northern Europe, affecting around one in every 2500 births, with a carrier frequency of about one in 25. Couples who are both carriers for the defective gene are themselves healthy, but have a one in four chance of having a baby affected by the disorder (Passarge, 1995). The isolation of the gene in 1989 was a major breakthrough, greatly enhancing the accuracy of diagnosis for affected children and carriers of the defective gene (Passarge, 1995).

The intention to offer testing to the public by the laboratory concerned had been announced in a press release the previous December (University Diagnostics, 1994), to the surprise and consternation of health professionals already involved in extensive evaluation of the possible advantages and disadvantages of offering carrier screening for cystic fibrosis within the primary care setting (Harper, 1995). That a commercial development would seemingly overtake the UK genetic centres to market, without consultation, a service directly to the public, had seemed improbable.

The subsequent debate has centred as much on the implications for the future of genetic testing as on the issues that have arisen from the initiative itself. The Government has come under increasing pressure to provide guidance for providers and ‘consumers’ of genetic tests, finally responding in early 1996 by appointing the Advisory Committee on Genetic Testing (ACGT) to advise the Departments of Health and Trade and Industry. Its Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public was published last month (ACGT, 1997).
What is the problem?
It could be argued that any private initiative that widens public access to healthcare services is to be welcomed. Further, that the provision of genetic tests is simply an extension of the current range of tests already available to the public, such as kits to test for pregnancy or measure cholesterol levels, or even provision of services in local chemist outlets to measure blood pressure. Does the availability of a test to ascertain carrier status of a genetic disorder really potentiate any different problems from other self-diagnostic kits such as these? In any case, is not the uptake of such tests likely to be small? What are the implications for the future of genetic testing? This article sets out to review some of the current concerns about offering genetic testing directly to the public, and aims to clarify some of the issues of which the nursing profession should be aware, not only as healthcare professionals, but also as members of society.

Gene testing: current status
Although there is currently only one gene test on offer commercially in the UK, a second laboratory now offers this test along with antenatal screening for Down’s syndrome (The Times, 1997) and other commercial laboratories have announced their intentions to offer genetic tests in the future (Bird, 1997). The concern is that there is a vast potential for the market to be expanded to incorporate tests for a wide range of human conditions before the impact on the individual and the implications for society have been fully evaluated. This potential increase is as a result of progress in molecular technology and genetics research, dominated by one project in particular: the Human Genome Project is an international multi-million pound project to map and sequence the entire human genome. It is estimated that this will take 15 years; the first 5-year stage was completed successfully in 1995 (Passarge, 1995).

It is widely accepted that this major project, along with associated research efforts, will have significant implications for public health and clinical medicine. Once the gene itself has been identified, then a diagnostic test can be developed to detect the defective gene in an individual. In some cases, identification of a gene for a specific disorder has led to the availability of clinical testing within months of discovery. It is thought that all major disease susceptibility genes will be identified in the next 5-10 years (Genetic Interest Group, 1995). As a consequence, tests for inherited monogenetic disorders such as cystic fibrosis and Huntington’s disease, and tests for genetic predisposition to common diseases such as diabetes, schizophrenia, cardiovascular disease, and a wide range of cancers, will be developed; therefore, an increasing number and range of new tests based on genetics research can be anticipated.

Potential uptake
While there appears to be a consensus that opportunities for genetic testing will increase, public demand for testing is uncertain, and the Welsh Health Planning Forum (1995) feels demand should not be over-estimated. There is, as yet, only limited indication of the likely public response. Since its advertisement, University Diagnostics has indicated that, up until December 1996, between 500 and 1000 people have undergone testing. Recent research indicates that uptake of predictive testing for breast cancer would be high (Julian-Reynier et al, 1996; Laloo et al, 1996).

In their survey of the potential uptake of testing for mutations in genes that predispose to breast cancer (BRCA1), Laloo et al (1996) found that 93.75% of women asked from the general public would undergo predictive testing if it were available, as would 87% of women attending a breast clinic, and 91% of women attending a family history clinic for counselling. Tests for mutations in the BRCA1 gene are available commercially in the USA, with discounts offered for family testing.
Willingness to pay for testing has not been fully ascertained in the UK. Miedzybrodzka et al (1995) found that the only significant predictor of willingness to pay for carrier screening for cystic fibrosis by women attending antenatal clinic was social class.

**Why is it different?**
Genetic tests differ from other clinical tests because the information revealed may have implications for the family as well as the individual; it is shared information and, as such, poses unique challenges to confidentiality and privacy of information (Boddington, 1994). Individuals may wish to gain information about another family member in order to clarify their own genetic status: this may conflict with the desire of that family member not to be involved in testing. Similarly, information about an individual may automatically reveal information about a close relative who chooses not to know his/her genetic status. Non-paternity is another issue that could create potential difficulties.

Another distinguishing feature of genetic tests is that they may predict the potential future health of an individual, and at a much earlier stage in development that other medical screening tests. An individual may be tested positive when pre-symptomatic for a disease of late onset such as Huntington’s disease, or testing may indicate predisposition to future disease symptoms. This may have implications for employment and insurance as well as for the wellbeing of the individual and families concerned. The importance of counselling as an integral part of testing in such circumstances has been emphasized (Harper, 1995).

Genetic testing for asymptomatic individuals, whether due to carrier status of a recessive disorder, or to late-onset disorder, may provoke anxiety in otherwise healthy individuals. It has also been identified that simply offering a test could raise anxiety, and testing itself may lead to an increase in psychological problems and disturbed family relationships (Welsh Health Planning Forum, 1995).

Testing to determine the genetic status of an individual is clearly not straightforward; it raises potential ethical and psychological issues not commonly encountered in other clinical diagnostic tests. As such, genetic testing places an additional onus on those who offer testing, if individuals and their families are to benefit from testing, and furthermore, is they are not to be harmed by testing.

**Implications of commercial gene testing**

*The counselling process*

While it is agreed that genetic testing, such as carrier screening for cystic fibrosis, can be undertaken without causing harm, in studies indicating this, the diagnostic test itself formed only part of a process of genetic counselling by skilled professionals. The provision of information and support, and how it is delivered, is a significant contributory factor to avoiding harm from receiving genetic information (Michie et al, 1996).

Skirton (1994) emphasizes the importance of the counselling role within the provision of genetic services. Gene testing, particularly by ‘mail order’, runs a serious risk of being seen in isolation from the counselling process when it is offered as a diagnostic service. Counselling itself, by skilled professionals, is costly and time-consuming, with suitable outcome measures being difficult to identify and assess with the current framework of NHS service provision (Kirk, 1996). The extent to which commercial testing houses would offer accessible counselling for all of their customers, both before and after testing, and irrespective of the test result, needs to be clarified.
Understanding the test results
One important aspect of information provision is the client’s understanding of the implications of the result, irrespective of the outcome of the test. In the case of negative results, it may not be fully appreciated that such and outcome does not necessarily indicate unequivocal ‘good news’ with no further risk of disease or anxiety. Three issues may be considered to illustrate this:

- Not all possible mutations within a gene may have been tested, such as might be the case for cystic fibrosis. Thus, although an individual may receive a negative result for the common mutations associated with the cystic fibrosis gene, there is nonetheless a residual risk for the rarer mutations not tested for.

- False assurance may be given by a negative result for an inherited disorder, such as familial breast cancer, where the individual would still be left with the general population risk of developing non-inheritable breast cancer. Breast cancer can be due to the cancer predisposition gene BRCA1 probably accounts for about 2% of all cases of breast cancer (Eeles, 1996). Individuals testing negative would need to understand that they should continue with surveillance such as self-examination.

- A personal account provides a powerful illustration of the need for testing to be seen as part of a process of counselling, accepting that an individual receiving a negative result may well require further support (Madigan, 1996). Although tested negative for Huntington’s disease, Julia Madigan was still receiving counselling support 4 months after receiving her ‘good news’. She writes of the mixture of feelings experienced - relief, happiness, guilt, isolation (from affected family members) and deep depression - as she tries to adjust to the implications of the result. The issue of counselling after a negative result will be particularly relevant when other family members may have tested positive for a disorder.

The implications of a positive result can also present difficulties in understanding. The outcome may represent a risk, not a certainty, and this may be further complicated by variations in severity of symptoms manifested, depending on interactions with other genes, and with environmental factors. Davison (1996) points out that even when a defective gene is identified in an individual, the expected disorder may never be manifest, and in most circumstances, the time of onset of symptoms will be unknown. A client testing positive for the BRCA1 mutation, for example, faces a 51% risk of developing the disease by 50 years, and an 85% risk of developing the disease by the age of 70 years (Eeles, 1996).

The result of a genetic test, whether it is positive or negative, can generate ambiguous or complicated information that requires clear explanation. Client understanding needs to be ascertained, and it is difficult to see how provision of information by post, or by any other form of non-direct contact, could provide opportunities to test this. Provision of a customer helpline would not necessarily address this issue. Eeles (1996) asserts that test results should be given only by those who understand them, and feels that gene tests should not be sold direct to the public.

Implications for individuals and society
It has already been mentioned that the outcome of a gene test may have implications for insurance and employment. The potential for discrimination against those who have undergone, and been tested positive for, a predictive genetic test is worrying, and clients should be aware of this before agreeing to a test. Although there may be sound commercial reasons for insurers to assess the potential future health of prospective clients before they set premiums, and to adjust or even exclude potential ‘high-risk’ clients to the benefit of ‘low-risk’ customers, as a society we should question the acceptability of such practice.

It has been argued that society should preserve the principle of provision of care irrespective of circumstances, and should agree to forgo any premium advantage to being able to show when
individuals are genetically low-risk (The Lancet, 1996). Whether society would agree to this is uncertain, but there is a clearly a need for this issue to be widely debated and resolved.

A second area of concern is that gene testing by mail-order would appear to offer little provision to safeguard against inappropriate testing of children. The Clinical Genetics Society in the UK currently advises against children undergoing carrier testing or predictive testing for disorders of late onset, and the issues around this are subject to ongoing research (Clarke and Flinter, 1996). How commercial testing laboratories could protect against this while samples are not collected in the presence of staff is unclear. The extent to which the voluntary Code of Practice will address this difficult area should be monitored closely.

The positive impact that gene testing can bring to individuals should also be considered, albeit with the proviso that the testing process provides information to individuals in such a way that they are able to benefit from it. Predictive testing may offer options for early treatment, or preventive adaptations such as altering lifestyle. It also may assist couples in making decisions about unborn or future children, and to make plans and provision for the future. However, in this context we may wish to question whether such testing should be offered as a service, rather than through private initiative for commercial profit.

Implications for nurses
Despite being difficult to assess, the indications are that there are significant deficits in public understanding of genetics, although interest in the area is growing (Durant et al, 1996). The advent of wider opportunity for gene testing via commercial venture can only contribute further to public interest and awareness, and perhaps anxiety. It would be reasonable to suppose that in wishing to gain further information and clarification of issues arising from this, members of the public should seek the views and advice of nurses, particularly those in the primary care sector. If nurses are to address this demand, there are immediate implications for their own levels of awareness and understanding of concerns about gene testing in general, and commercial ventures in particular:

• Nurses need to be aware that gene tests are currently available direct to the public, and that the range and availability of such tests is likely to increase.
• Nurses need to be aware that there are advantages and disadvantages to undergoing gene testing; if clients are to give truly informed consent to a gene test, they need to be aware of these. A client who is pregnant, or who has a current family history of a genetic disorder, will be particularly vulnerable to potential harm from an inadequate counselling process.
• Nurses need to know where they, and their clients, can seek further information and advice. Thus, they need to be familiar with local provision of genetic counselling services by the regional genetic centres.
• A health educators, nurses should be willing to promote education and understanding if gene testing. One way in which we may contribute to understanding is by participation in public and professional debate, offering informed opinions that incorporate nursing knowledge.

There is another consideration for nurses in assisting the public ‘through the genetics maze’. Genetics has been gaining an increasingly higher profile in the media, and any advertising campaign for commercial testing may only add to the hype. We need to maintain a perspective on the importance of genetics in health care and present a clear vision of the realities offered by genetics advances, discarding the fiction. At the same time, we need to ensure that other healthcare issues are given the profile they deserve in relation to health gain.

This approach requires an education that, while providing a fundamental understanding of the scientific basis of inheritance, genetic disease and molecular genetics, does not present this as the prime focus for genetics teaching. Rather, the emphasis on genetics in a nursing curriculum should
be on the impact it has in relation to health care, both currently and potentially, on individuals, their families, and society as a whole, and on the decisions that we all have to make in relation to this.

Conclusions
Although the ACGT has now published its first report on genetic testing services offered direct to the public, the future possibilities offered by such testing, and their impact on health care, remain uncertain. Issues surrounding commercial gene testing have also to be considered within the wider context of the rapid progress in genetics research and technology, and its impacts on healthcare provision and services. While public and professional appreciation of the implications of such advances struggles to keep pace, we are at risk of allowing technology to lead demand for genetic information and services. Until all the implications of gene testing have been debated by an informed public and health professionals, the author feels that gene tests should not be offered commercially direct to the public.

Key Points
• Gene testing has been offered commercially in the UK since 1995; the range of tests offered directly to the public is likely to increase.
• Gene testing presents its own unique benefits and problems to individuals and society.
• While awareness is increasing, the full implications of the issues of gene testing have yet to be appreciated by both the public and health professionals alike.
• Nurses should play a key role in participating in the ongoing debate, informing the public, and maintaining a perspective on the issues surrounding gene testing.
• Until the implications of gene testing have been debated by an informed public and professionals, the author feels that gene tests should not be offered commercially direct to the public.

Disclaimer
Any views expressed by the author do not necessarily reflect those of the Genomics Policy Unit.

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