Let the consumer decide? The regulation of commercial genetic testing

Dr Mairi Levitt

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Dr Mairi Levitt *University of Central Lancashire, Preston*

Abstract

Objectives—*The development of predictive genetic tests provides a new area where consumers can gain knowledge of their health status and commercial opportunities. "Over-the-counter" or mail order genetic tests are most likely to provide information on carrier status or the risk of developing a multifactorial disease. The paper considers the social and ethical implications of individuals purchasing genetic tests and whether genetic information is different from other types of health information which individuals can obtain for themselves.*

Design—*The discussion is illustrated by findings from a questionnaire survey of university students as potential consumers. Topics covered included what health tests they had already used, expectations of genetic tests, willingness to pay, who should have access to the results and whether there need to be restrictions on such tests.*

Sample—*Six hundred and fifteen first-year students in the universities of Leuven, Cardiff, Central Lancashire, Vienna and Nijmegen studying either medicine or a non-science subject.*

Results—*Students were enthusiastic about genetic tests and had high expectations of their accuracy and usefulness but most thought they should be available through the health service and a minority thought that some tests, for example for sex selection, should not be available at all. There were few differences in responses by sex or subject of study but some by country. The paper also considers ethical and social issues outside the scope of a questionnaire survey of this type.*

Conclusion—*To address some of these issues the sale of genetic tests to individuals can be made subject to ethical guidelines or codes of practice, for example to protect vulnerable groups, but there are fundamental social and ethical questions which such guidelines cannot address.*

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Keywords: Genetic tests; European; commercialisation

Health as lifestyle

Health education has made people increasingly aware of all kinds of health risks. Lifestyles, food, drinks, additives and chemicals are risk factors given media attention. Students, in the study discussed below, were asked to rate a list of factors in terms of their importance for maintaining health. Three of the first four factors they chose were things over which the individual might be thought to have control: diet, exercise and type of work, the fourth was infections. The individual expects, and is expected, to be proactive in health lifestyle in contrast to a fatalistic view of health and illness. It might seem that genetic makeup is outside the person's control but knowing your genetic risk is becoming another task for the responsible individual. The focus on individual responsibility and control over health provides the right social climate for "over the counter" testing as a way of exercising this responsibility as a rational and autonomous individual.

A study of potential users

Discussion of some of the ethical and social issues raised by over the counter tests will be illustrated by data from a pilot study carried out among a group of potential personal users. A questionnaire study of attitudes cannot, however, cover all the ethical and social issues, particularly those looking at fundamental questions about priorities in health care and the distribution of limited resources. The study used a sample of students who were chosen as likely to be among the first users of genetic test kits because of their future occupations and earning capacity. It was decided to give out questionnaires to a whole first-year group taking medicine and another group taking a non-medical, non-science subject. Six hundred and fifteen university students taking first-year courses completed questionnaires in English, which were given out to all students attending a lecture. The questions were mainly closed, with space for comments. Forty-four per cent were medical students and the rest were studying philosophy, law or social science. The majority were from Belgium and the UK, with a smaller number from the Netherlands and Austria.[1] Just over half were female and 90 per cent were in the age group 18-25 years. The reason for the high numbers in Belgium was that the response rate was 100 per cent because the questionnaires were given out and taken in during a large first-year lecture course. In the other countries the response rate was around a third because students were given the questionnaires during a lecture and asked to bring them back. The responses may not therefore be typical even of the full student group, except in Belgium. The data are simply used, however, to provide comment on issues raised by genetic testing and it was found that on most issues there was agreement between countries, between men and women and between medical and non-medical students.

Summary of questions

Students were presented with two groups of statements giving different opinions on genetic research and testing and asked whether they agreed or disagreed with each (16 statements in all). These were balanced in terms of enthusiasm for and reservations about testing; for example: "I want to know about testing for my partner and me, to see if our children might be born with a genetic disorder" and "I would only want to be tested for genetic diseases which can be treated". Belgium and the UK gave similar answers and were enthusiastic about testing. For example, "I want to know as much as possible about any genetic disease I might develop": 69 per cent of Belgium and 71 per cent of UK students agreed, with no sex difference. However, the percentage agreeing that "there is too much emphasis on genes as causes of disease rather than the environment in which we live" was 57 per cent overall, highest in the UK at 62 per cent. Students were given a list of health care tests including genetic tests and asked if they had had the test, whether they had bought it themselves, might be interested in buying it, were not interested in it or had never heard of it. They were also asked where these tests should be available (for example, for individuals to buy, through the health service/family doctor, or not at all). They were asked whether they would be willing to pay for a test for a breast cancer mutation for themselves; whether they would encourage their partner to do so; the maximum amount they would pay; whether they would pay extra for counselling, and to give the reasons for their answers in open questions. Students were asked whether they thought there needed to be restrictions on genetic testing and, if so, what they should be; a specific question on the genetic testing of currently healthy children was included. Students were asked whether a person with an unfavourable test result had a duty to tell anyone. They were asked to rate their concerns about tests provided by their national health service and by private companies. It was thought students might be much more concerned about the quality of tests provided by private companies as opposed to their national health service, but this was only evident in the case of safeguards for children when tests were provided privately, and among the UK sample, who were generally more concerned about non-National Health Service (NHS) tests. Students were concerned about accuracy, confidentiality, the availability of advice and that the limitations of tests should be explained. A separate question asked how accurate they would expect different types of genetic tests to be, ie for carrier status (term explained), for a late onset disorder, and prenatal screening.

Commercial health tests

Individuals in Britain can already buy health tests to measure blood pressure, cholesterol, pregnancy, diabetes and fertility. Mail order genetic tests include carrier status for cystic fibrosis and a paternity test which uses a cheek swab from father and child. Newspaper advertisements for the paternity test from the DNA testing agency, Keston, Kent, read: "Whose child is it? Are you really the father?"[2] In the US tests marketed directly to the public include tests for susceptibility genes for breast and ovarian cancer (BRCA1 and 2) and genetic predisposition to cardiovascular disease; these are availalbe from from Myriad Genetics, Salt Lake City. A test for HIV is available from Home Access Health Corporation.[3] Small numbers of students in the sample had bought tests for cholesterol level, blood pressure and pregnancy but more had been tested for the first two within the health service. The few who had taken genetic tests had been tested within their national health service, most for cystic fibrosis carrier status (three per cent of the sample).

It is difficult to estimate demand when the question is hypothetical but a majority of students reported that they would be willing to pay for a test for a breast cancer mutation or would encourage their partner to do so: 66 per cent of men and 62 per cent of women. Most would only be willing to pay small amounts, 57 per cent named an amount under 50 euros ($US 44), but for some this reflected a lack of money rather than a low priority for tests. One student wrote: "I would have to decide whether to eat or buy the test", while another would pay "whatever it costs" because "health has no price". In an open question most of those wanting the test said it was "best to know" so that they would know how to avoid risks and have a better chance of treatment or a cure. Breast cancer was chosen as an example of a well-known multifactorial disease but students may well have overestimated the genetic component. Overall 15 per cent wrote to the effect that it was "best not to know". Most of those answering would be willing to pay extra for counselling if the test result was positive but a substantial minority would not, from 25 per cent in Belgium to 47 per cent in Austria. The Austrian sample were on average older than the Belgium sample and fewer older people as well as fewer women were prepared to pay extra for counselling, and the amounts they were willing to pay were smaller. Overall 51 per cent were willing to pay 40 euros ($US35) or less for counselling. The benefits expected (by 91 per cent of those answering) were more information, a risk analysis, advice on the best course of action and/or mental support. Responses, included, for example: "Information about the test and the best course of action"; "a clear summary of results, relief and guidance".

Are genetic tests different?

It can be argued that there are no concerns, or no new concerns, surrounding genetic tests as opposed to other types of over the counter tests. In support of this view the following arguments can be made:

- Why should we be concerned about genetic tests when other self tests are available without regulation? As the president of Myriad Genetics's laboratory said: "there's really no difference between a blood cholesterol test and a genetic test. No one has given me a good reason why this type of test

would require FDA oversight, [in the states] whereas others do not."[4]

- Commercial companies will only market tests which people want to buy, if there is no market for them they will not be produced. People should be free to make up their own minds. As a spokesman for the DNA Testing Agency is reported to have said: "We are giving people the opportunity to do private testing...".[5]
- Commercial testing can fill gaps in provision which cannot be met by public funding. The European Diagnostic Association, commenting on the ethical issues raised in the draft report on this research wrote: "Industry believes it has an increasingly important role in delivering health care products and to a certain extent services where public funding is not available or is inadequate to meet patient demand".
- It is important for individuals/families to have the opportunity to be informed. The Genetic Interest Group argues that genetic services offer people "the potential to acquire information about their genetic make up ... though it may be bad news, allows them to plan out their lives and make informed reproductive decisions".[6] The group criticises the cautious approach to genetic testing of children in the Clinical Genetics Society report as "overly concerned with psychological consequences".[7]
- There are always people who are frightened by new technology, fuelled by media hype, but soon it becomes routine and accepted like test tube babies born through IVF. This point was made in the focus groups on cloning run by the Wellcome Trust.[8]

These arguments will be considered in more detail and illustrated, where relevant, with information from the students' responses to the questionnaire.

WHY SHOULD WE BE CONCERNED ABOUT GENETIC TESTS WHEN OTHER SELF TESTS ARE AVAILABLE WITHOUT REGULATION?

This raises the issue of whether genetic technology and the information it reveals is different from other health technology and information, and different in such a way that it has to be surrounded by special restrictions to protect the public. It is necessary to distinguish between types of genetic test. Diagnostic tests will make a definite explanation of a current condition, when the individual is already displaying symptoms, or of carrier status for a recessive disorder such as cystic fibrosis (although in the case of cystic fibrosis a test will be for the most common mutations so can never achieve a 100 per cent exclusion of a carrier status). Other tests will predict a future condition before any symptoms appear. It can be predicted that a newborn boy will develop Duchenne muscular dystrophy and will begin to show the symptoms in infancy. Genetic tests may reveal a predisposition/susceptibility to a disorder and can be used to calculate the risk of developing breast or ovarian cancer, cardiovascular disease or Alzheimer's. There are more commercial possibilities in tests for multifactorial conditions such as these because they will be more common and there is the potential to develop drugs to treat the conditions. It is the treatments rather than the test kits, which will generate profits.

Among these different types of tests those which diagnose an existing condition or carrier status may seem least problematic, but when offered over the counter there is no control over who is being tested or that person's particular situation. A parent might test a child or a pregnant woman might test herself and her partner. Samples may be collected secretly and sent for testing without the subject's knowledge, for example, hair collected from a hairbrush as a source for paternity testing. The same test will have different implications depending on the circumstances. In the case of child testing it takes away the child's right not to know about late onset disorders such as diabetes. To ask why genetic tests need regulation when cholesterol testing does not implies that both are unproblematic, but perhaps there should be more concern about cholesterol testing, particularly in children, rather than less concern about genetic tests.[9]

Two-thirds of the students wanted to know about any genetic disorder they, their partner or unborn children might develop but were less sure about testing children for a disorder they might develop later, with only one third in favour. The students from the Netherlands were consistently against parents testing their children. Students were divided 50:50 on whether they would want to be tested for untreatable disorders. They were asked if any groups needed special protection when genetic tests could be purchased by individuals and 96 per cent chose at least one group. The top four groups needing protection in order of choice were the same for medical and non-medical students in all countries:

1. Employees from employers: 69%;
2. Mentally handicapped people whose relatives want them tested: 66%;
3. Unborn children: 61%, and
4. Children and adolescents whose parents want to have them tested: 61%.

Students were asked what sort of protection would be needed for the groups they had selected and then to rate what, in their opinion, were the most important forms of protection. The top two choices in each country were that tests should be available only through health professionals and there should be government regulation; these were closely followed by European regulation. Although wanting information for themselves students saw the need for protection when those involved were not of equal power and status: employees and employers, mentally handicapped adults and their relatives, children and parents.

COMMERCIAL COMPANIES WILL ONLY MARKET TESTS WHICH PEOPLE WANT TO BUY

Commercial companies know that demand can be generated but the consequences may be more serious than generating a market for Teletubbies or designer trainers. Demand can be manipulated

through the way information is offered. If a test is simply carried out routinely, like the phenylketonuria (PKU) test for newborn babies, then an uptake of around 100 per cent can be achieved. Uptake can be gradually reduced by putting extra steps or obstacles on the path to a test, for example, a card to be returned before an appointment is made, pretest counselling and a waiting period. This was illustrated in a pilot screening programme among newborn boys in Wales for Duchenne muscular dystrophy.[10] The type of information with which potential patients are provided is also important. Information on new tests for the general public and in the media tends to be positive, reflecting the views of the researchers and making claims about improved treatment or cures "soon". However, the Huntington's gene discovered in 1993 has not yet led to better treatment and current treatment for sickle cell anaemia was developed before the gene was identified. Multiplex testing, which offers packages of tests for different kinds of conditions in a single session, will further complicate the giving of pretest information and the obtaining of informed consent.[11]

The emphasis is on individual choice and informed consent but the most important variable in take-up rates for genetic testing is the way it is offered. The control offered to the public is in any case limited: they can take or reject a test but they cannot decide what test services ought to be developed in the first place. The public are not expected to argue that some tests should not be available at all.

COMMERCIAL TESTING CAN FILL GAPS IN PROVISION WHICH CANNOT BE MET BY PUBLIC FUNDING

It is the view of the industry that commercial testing can fill gaps in provision which cannot be met by public funding. A spokesman from a company marketing cystic fibrosis testing kits by mail order said: "There will be a growing demand from people wanting to learn about their genetic make-up. The NHS is already overstretched and GPs are not necessarily experts in genetics, the private sector will siphon off demand".[12]

There is concern that there will be greater health inequality if some tests with health benefits are only available to those who can pay. An alternative view is that tests may be offered which are not medically indicated, have no therapeutic or preventive options, do not test for a serious disorder and do not offer adequate certainty, in which case those who cannot afford them could be better off! Either way, commercial testing is likely to put a burden on the health system rather than fill the gaps because the test is only the beginning. If tests are available over the counter public awareness of tests will increase along with demand for them, whether they are beneficial or not. Once people have had a genetic test the NHS may be left to pick up the pieces. In the UK patients will probably go straight to their general practitioner (GP) for more information, as 93 per cent of UK students in the sample wrote that they would. It is likely that they would be referred to their regional genetics centre and perhaps retested if the status of the first test was uncertain.[13] Individuals, including children, will present who would not have been tested for the specific condition in the NHS or who would have decided not to be tested if they had received pretest counselling. Even those with a favourable result may seek a second opinion from a trusted source.

The gap in provision referred to by the industry may therefore be there for valid reasons. At the moment resources are concentrated on genetic testing for specific groups, for example, those with a family history of a serious genetic disorder who request preimplantation or prenatal testing, or where diagnosis would aid treatment. There are gaps in health provision which would benefit more people's health if they could be filled than extending genetic testing, for example social and environmental measures to improve housing conditions and to relieve child poverty. Although this argument could equally be applied to other medical procedures, such as heart transplants, commercial genetic testing is less likely to be a matter of life and death and more likely to be another health check for the affluent, the worried and the hypochondriac.

IT IS IMPORTANT FOR INDIVIDUALS/FAMILIES TO HAVE THE OPPORTUNITY TO BE INFORMED

There is an assumption that information is empowering but to know you have a late onset untreatable condition or a predisposition to a condition where treatment is uncertain may not be. Ability to predict a disease long before there are any symptoms is what is attractive about the technology but it creates a group of people who are "in an ambiguous position between health and disease" with implications for their life chances, including their insurability and employability.[14] Students supported choice in theory but when asked about specific tests most did not support over the counter testing for everything and a minority rejected some tests altogether. While 90 per cent agreed that "people should be free to make up their own minds about having a genetic test", 82 per cent supported at least some tests being available only through the health service and 35 per cent felt that a test to find the sex of an unborn child should not be available at all. While the argument stresses the "opportunity" to be informed, it may be difficult to reject testing, especially in pregnancy, or information may be forced on someone by the actions of other family members. "If everyone is getting tested then I have to be tested too".[15]

There is also the problem of the quality of the information received. Despite the information given in the questionnaire that most tests for carrier status "are not 100% accurate, for example in the test for cystic fibrosis carriers, only the most common mutations are included", a quarter of students expected that a test "would detect 100% of affected individuals". The more serious the implications of the test, the more accurate students expected it to be. Nearly half the students (46%) expected a test to see if their unborn child would

have a genetic disorder to be completely accurate. It is likely that counselling services would be available with commercial tests but these are less likely to be compulsory before the test will be given, for example, a telephone advice line and written information may be on offer rather than a compulsory pretest meeting with a genetic counsellor. While people may be attracted by the privacy of the tests this does not mean they are prepared for a "bad" result or for information that is proved later to have been inaccurate.

NEW TECHNOLOGY SOON BECOMES ROUTINISED

Tests for Down's syndrome are routinely offered to older expectant mothers but this does not mean they are now uncontroversial. Chris Goodey and colleagues commented in the *Bulletin of Medical Ethics* that the medical press concentrates on the techniques of screening, which diverts attention from the question "why screen for Down's syndrome, on what evidence and logic are the arguments which support screening based" (for example assumptions that Down's syndrome involves suffering).[16] Technology may become routine but this does not necessarily mean the ethical and social issues have disappeared or been resolved.

Acceptable and unacceptable practices are assumed to be easily distinguishable and supporters of testing pick on clear-cut examples to make their point. For example, the director of the Genetic Interest Group wrote: "Pro-life groups believe that the increased use of antenatal genetic testing will lead to a dramatic increase in the number of abortions carried out for trivial imperfections, such as hair or eye colour".[17] This invites the (presumably intended) response: "how ridiculous, no one would do that", particularly as pro-life groups are seen by many in the UK as extremists. However, the public recognise the existence of a large "gray area" around research and practice and the difficulties of "drawing the line".[18] While there is no problem in "drawing the line" at aborting for eye and hair colour what about cystic fibrosis, cleft palate or achondroplasia?

Professional and expert discourse in the new genetics

The rhetoric surrounding the use of genetic technology in health is that of individual choice or consent from an informed position. Promoters of commercial screening use this rhetoric to argue that over the counter and mail order gene tests add to the choices for individuals and families. The stress on choice is used to deflect criticism of social engineering and eugenics because the public, not the state, will be in control. When I met with three representatives of the diagnostic industry to present the survey findings, they emphasised their role as offering the opportunity for testing but said that it was up to society to decide whether tests should be available to the public. In fact, however, the diagnostic industry has a major influence on policy in the UK. The industry's representative on the advisory committee on genetic testing was one of the four members of a subgroup which produced the *Code of Practice and Guidance on Human Genetic Testing Services Supplied Directly to the Public*.[19]

This code of practice addresses social and ethical issues and recommends, for example, that tests should not be supplied to those under 16, and that suppliers should provide "full information" and opportunities for "pre and post test genetic consultation [with] no additional charge" and should obtain written consent to supply a copy of test results to the customer's GP.[20] Genetic tests can be said to be subject to a code of practice and fears of unrestrained commercial testing allayed. Thus individual tests gain an ethical gloss while the more fundamental questions of the commercialisation of genetic testing have not been tackled. The questions to be debated include: how far "drawing the line in genetic testing" can be left to individual "choice"; what constitutes "normality" and suffering; how the availability of a test for a condition might affect those who have the condition, and how far people can be said to make a choice when they are dependent on expert information and subject to social pressures.

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Dr Mairi Levitt, BA, MA, Dip Ed, PhD, is Senior Lecturer, Centre for Professional Ethics, University of Central Lancashire, Preston.

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