



Human
Genetics
Commission

The supply of genetic tests direct to the public

A consultation document

July 2002

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An HGC Consultation Document

The Human Genetics Commission (HGC) is an independent body that advises the Government on the impact of developments in human genetics, in particular on the legal, ethical and social implications.

In May this year the Commission published a report - Inside Information – which made a number of recommendations about genetic information, and how genetic information should be given to people in a clinical context. The Government has now asked HGC to examine the issue of genetic tests which are offered directly to the public, outside of the normal process of referral by a medical practitioner. The Commission aims to publish its advice to Government by the end of 2002.

A Working Group of the HGC has drawn up this consultation document which explains the scope of this review, summarises the issues and poses a number of questions about the existing controls, possible future developments and the position in other countries or in comparable areas of health and consumer protection.

HGC would welcome responses to any or all of these questions or any related matters that you wish to draw to our attention. We would like to publish the responses. Please let us know if you want part or all of your response to be treated in confidence.

Please send your comments to:

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All responses should arrive by Friday 4 October 2002

Introduction

1. Developments in human genetics have brought an increased understanding of genes and of what they do. One effect of this has been to increase the number of genetic factors that can be tested for. Until recently, genetic tests were available for only a handful of inherited conditions, now they are available for a broad range of health risks and medical conditions. These tests may do a variety of different things:

- diagnose disease that is already present or which is likely to occur in the future;
- detect carrier status in an individual who may not be affected by a condition but who may pass it on to descendants;
- disclose the presence of gene variations that increase the risk that a person will develop a complex condition such as cancer or heart disease;
- determine reactions to medicines, chemicals, foods or environmental factors; or
- establish family relationships (paternity testing) or historical roots (genealogy testing).

In short, these tests are capable of revealing important information about the past, current and future health of the person tested and sometimes give information about their relatives.

2. Genetic testing has mainly been offered within the context of medical diagnosis and treatment as a specialist discipline within the National Health Service (NHS). Typically, a genetic test is initiated by a general practitioner (GP) who makes a referral to a specialist such as a clinical geneticist or other consultant. He or she undertakes the testing after discussing the situation with the patient or, where relevant, the parents of a child. Genetic testing has tended to be seen as a specialist service, provided under medical control.

3. Two factors could change this picture of genetic testing in the UK. Firstly, the technology of genetic testing has become simpler and more readily available, thus making rapid and relatively inexpensive genetic testing a reality. Secondly, this development has been accompanied by a growing understanding by the public of medical information and a corresponding desire to make health decisions on the basis of information directly acquired and understood by the patient without the involvement of an intermediary such as a doctor. This shift to personal responsibility and partnership in health decisions has been encouraged by governments and is consistent with the vision of better preventive medicine. Finally,

individuals may want to access such services to avoid delays or difficulties in accessing NHS services.

4. Some commercial companies are responding to an increasingly informed and interested public by directly marketing genetic testing products or services. This can be done through shops, for example, or by post. Such companies regard themselves as doing no more than providing information which the individual has the right to receive: information about his or her own genetic characteristics and the implications that these may have for decisions about health and lifestyle.

5. Against that might be balanced a recognition of the need to protect individuals from being given genetic information for which they are not prepared and which may lead to needless anxiety or false optimism. This in turn may lead to increased pressure on GPs and other NHS resources for referral and re-testing.

6. These are the issues that HGC is now addressing and on which this consultation focuses. We are therefore seeking views on whether there should be regulation of such “over-the-counter” testing, and, if so, how might this be achieved? Once we have completed our consultation we intend to issue advice to Ministers by the end of 2002.

The current legal position

7. At present genetic testing services supplied direct to the public are regulated by a mixture of specific and non-specific regulation and voluntary compliance mechanisms. There are no specific legal requirements in the UK about who may supply tests or to require them to comply with any laboratory standards or any protocols in disclosing results to the client. No professional qualifications are needed to obtain samples, subject them to analysis, and communicate the results to the client. Before seeking views on whether the current framework is adequate, we review in more detail some of the existing controls below.

Code of Practice

8. In 1997 the then Advisory Committee on Genetic Testing (ACGT; which was subsumed into HGC) published a *Code of Practice and Guidance on Human Genetic Testing Services*

*Supplied Direct to the Public*¹. This is a voluntary code and there are no legal sanctions attached to a failure to comply with it. At the time of its publication, it was thought that the main demand for over-the-counter genetic testing would be for tests to determine carrier status for inherited recessive disorders (such as cystic fibrosis). These conditions have no known health implications for the carrier. It was not envisaged that there would be interest in offering genetic tests for inherited dominant and X-linked conditions, or for certain adult-onset conditions, or for multifactorial or acquired disease. These latter tests were considered unlikely to be appropriate for over-the-counter testing either because they required family history information in order to be interpreted or because they were surrounded by uncertainties that required medical assessment. The Code was therefore intended to deal only with a very limited number of genetic tests, and it was anticipated that the Code would need to be reviewed regularly. The Code of Practice is ill-designed to deal with the wider range of genetic tests that might be supplied direct to the public.

9. There is also a Code of Practice on Genetic Paternity Testing Services², published in 2001, which sets out good practice for testing mothers, their children and putative fathers in order to establish parentage. Such tests generally involve a DNA test that is offered direct to the public, but we do not intend to deal with them at this stage of the review. This is a distinct area of testing which raises different issues, not least that it often involves the testing of third parties who are children. We feel that it is more appropriate to focus in this consultation on tests aimed at health and lifestyle issues. We will consider the subject of paternity testing at a later stage.

Regulations

10. There are regulations that set out standards for testing kits. The In-vitro Medical Device (IVD) Regulations (SI 2000 No 1315) require IVDs to meet safety, quality and performance requirements before they are placed on the market with a CE mark (see Annex B). These devices are regulated by the Medical Devices Agency. This system of regulation, however, is confined to the equipment and reagents used to test, and has no bearing on aspects of the testing service such as its appropriateness or medical value.

1. <http://www.doh.gov.uk/genetics/hgts.htm>

2. <http://www.doh.gov.uk/genetics/paternity.htm>

11. There are also specific regulations that control the sale of HIV testing kits direct to the public. The HIV Testing Kits and Services Regulations 1992 (and parallel Regulations in Northern Ireland):

- make it an offence to sell, supply or advertise for sale or supply an HIV testing kit or component to a member of the public in the UK;
- require the inclusion of specific warning notices in kits marketed in the UK about the need for confirmatory testing of a positive result and the possibility that recent infection with HIV may not be detected;
- make it an offence to provide HIV testing services that are not directed by a registered medical practitioner.

12. The regulations were introduced to maintain a system of free, anonymous (if desired) and confidential HIV testing for those concerned about their HIV status. It also ensures pre- and post-test face to face discussion with a trained health care professional including, arrangements for relevant follow-up. We consider later whether there are some types of genetic test that might merit similar regulatory controls.

Laboratory standards

13. At present there are a number of voluntary arrangements for ensuring laboratory standards in the NHS and private companies. Laboratories and companies are usually accredited to a recognised international quality standard such as ISO EN 17025 or ISO9002 or are accredited by Clinical Pathology Accreditation (UK) Ltd (CPA). This and similar bodies ensures that organisations meet the relevant standards covering organisational, professional and technical performance. These standards are internationally compliant, and systems of inspection exist to ensure that they are met by registered laboratories. Laboratories also need to participate in an established external quality assurance programme, of which there are several covering clinical genetics laboratories and paternity or forensic testing laboratories. There is, however, nothing to stop an unaccredited laboratory from offering genetic testing direct to the public, in which case the client would have no guarantee as to that laboratory's standards of performance.

Professional self-regulation

14. Many aspects of healthcare rely on professional self-regulation and require practitioners to be registered with a recognised body such as the General Medical Council, Pharmaceutical

Society, or the Nursing or Midwifery Council. There is also a new Health Professions Council (which replaced the Council for Professions Supplementary to Medicine from April 2002). It provides a multi-professional regulatory body that will deal with professional competence and lay input in some of the allied health professions relevant to this review, for example genetic counsellors, dieticians, clinical scientists and biomedical scientists.

15. There are in addition a variety of mechanisms to control practitioners in complementary and alternative therapies and products. The general approach towards self-regulation is to encourage practitioners to register with voluntary regulatory bodies, where they exist. These provide a variety of professional self-regulatory compliance and monitoring functions and a practitioner is free to practise without being registered with the relevant body. Only in certain circumstances where there are definite public health concerns has the Government introduced statutory controls. Osteopaths and chiropractors are regulated by the Osteopaths Act and Chiropractors Acts of 1993 and 1994 respectively. More recently, Health Ministers have given a commitment to bring acupuncturists and herbalists into statutory regulation. The coverage and content of statutory regulation for these professions has not yet been decided and will be the subject of public consultation in due course. In both cases, however, statutory regulation will be achieved by means of an Order made under Section 60 of the Health Act 1999.

Q1: What role, if any, do you envisage for professional bodies in the regulation of genetic testing services supplied direct to the public?

We consider later the possible self-regulatory role of industry bodies.

General consumer protection and other legislation

16. We must also recognise some of the broader UK and European legislation aimed at protecting consumers. There are a variety of relevant legal duties on suppliers of goods and services, which are enforced by local trading standards officers and the Office of Fair Trading (OFT). Of particular relevance to this review is the OFT's duty to encourage trade and professional bodies to prepare and disseminate information to protect consumers and to ensure that businesses operate fairly. In May 2002 the OFT published core criteria for

consumer Codes of Practice³ which must be met in order for any Code of Practice to be endorsed by the OFT. The aim of the core criteria is to ensure that a Code of Practice provides additional benefits and protections to consumers over and above the requirements of the law. The National Consumer Council, for example, has also published guidance on the principles to be considered in achieving effective voluntary self-regulation – so called soft law.

17. The Data Protection Act 1998 (DPA) is another example of general legislation that provides a number of safeguards of personal information that may be generated or stored as part of a genetic testing service. There are relevant issues of consent before data can be processed (which has a wide definition), the retention of data, and by implication samples and the processes that may be necessary to adequately anonymise data and samples (if that is an agreed part of the service). HGC has previously commented on the importance of providing unambiguous advice on the legal protection and duties under the DPA, and this might be achieved in any future Code of Practice.

18. We also note the differing legal position in Scotland. Of particular relevance are the crimes of reckless conduct and reckless injury. It is not necessary to prove that harm was caused to an individual, rather that there has been recklessness regarding injury to a member of the public. For example a company or person who shows recklessness in providing genetic information could be committing an offence.

Advertising

19. There are also self-regulatory controls on advertising in print, billboards, direct marketing, cinemas and the internet, overseen by the Advertising Standards Authority. The British Code of Advertising and Sales Promotion⁴ sets out a number of general principles – for example that advertisements must be legal, decent, honest and truthful and that adverts should be prepared with a sense of responsibility to consumers and to society. There are specific provisions relating to health and beauty products, including the provision of evidence for the claims made, preventing claims that might lead to misdiagnosis, or prevent medical treatment. In addition where adverts offer individual treatments, particularly those that are physically invasive, the advertiser may be asked to provide full details of the treatment and about those

3. Core criteria for consumer codes of practice. May 2002, available from <http://www.of.gov.uk>

4. <http://www.asa.org.uk> and http://www.itc.org.uk/itc_publications/codes_guidance/

who will supervise and administer them. This is backed up by the Control of Misleading Advertisements Regulations 1988 which allow an injunction to be sought to prevent misleading advertisements. There is also specific legislation regarding the advertising of medicines and medicinal products and corresponding codes produced by the pharmaceutical industry, which are perhaps somewhat outside of the scope of this review.

20. In addition, there are some specific prohibitions, such as the Cancer Act 1939, against the advertising of “...an offer to treat any person for cancer, or to prescribe any remedy therefor, or to give any advice in connection with the treatment thereof?”. These provisions have been used to prosecute companies selling devices or food supplements which they claim can prevent or cure cancer. The Cancer Act might also affect some genetic testing services if the advertiser claims to prevent or cure cancer.

21. The regulation of broadcast adverts is currently the responsibility of the Independent Television Commission (ITC) or the Radio Authority. We have not made a detailed examination of the various statutory requirements and Codes affecting broadcast adverts, but in general the restrictions are greater than for print advertisements. For instance, under the current ITC Code, television adverts may not contain any offer to diagnose, advise, prescribe or treat by correspondence. There are also clauses aimed at preventing erroneous self-diagnosis.

Q2: What aspects of the existing controls on advertising would you wish to draw to our attention in conducting this review? In what way, if any, should adverts for direct genetic testing services be treated differently?

Controls in other countries

22. We are concerned to consider the range of comparable controls in other countries, particularly those with a different system of healthcare provision. We are also aware of the importance of international instruments, particularly the European Union directives on goods and services. There are also several international treaties, conventions and declarations which apply, at a high level, certain duties regarding consent, confidentiality and the provision of adequate information. An example of this, the Council of Europe’s Convention on Human Rights and Biomedicine, Article 12 of which states:

“Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling.”

Work is underway on a protocol to this Convention which will provide further interpretation of the general Convention.

23. The Organisation for Economic Cooperation and Development (OECD) held a workshop on genetic testing⁵ in Vienna in February 2000, which summarised the legislation at national and international level. Similarly, the European Society of Human Genetics published a detailed report on genetic screening in July 2000, which considers the commercialisation of genetic testing and screening services.

24. One particular aspect of international controls which is relevant to this review is whether any international instruments exist which might limit the ability to promote or access certain types of genetic testing service via the Internet. A simple internet search for “DNA testing service” shows numerous entries, the majority of which are for paternity testing. A few hundred entries refer to other types of genetic testing services. Whilst it may appear that a number of companies offer genetic testing services, the majority of them, particularly in the United States, depend on a referral from a medical practitioner and will not conduct a test without proof of consent and adequate pre-test counselling. They often also rely primarily on health insurance policies to pay for the tests. Nevertheless, there are several companies in the UK and US that are, or are planning to, offer genetic testing services for a variety of conditions.

25. We shall be seeking information on comparable national controls as part of the review. The position in other countries is more complex than in the UK because of the different approach to the provision of healthcare and of predictive and diagnostic testing services by private companies. The Australian Law Reform Commission is currently conducting a major consultation on genetics and privacy, which considers direct genetic testing in passing. The United States has been actively considering the provision of genetic tests direct to the public since the ACGT Code of Practice was published. We have noted the conclusion of the 1997

5. <http://www.oecd.org/EN/document/0,,EN-document-617-1-no-20-3273-617,00.html>

US National Institutes of Health Task Force on Genetic Testing⁶ that advertising or marketing of predictive genetic tests to the public should be discouraged. In response to this report, the Secretary of Health and Human Services established the Secretary's Advisory Committee on Genetic Testing (SACGT) which recommended that genetic tests should be evaluated by the Food and Drug Administration before entering clinical service. An Interagency group has been established to consider laboratory standards, the analytical and clinical validity of tests and to conduct post-marketing data collection.

Q3: What other national controls or international instruments should we be considering during this review?

Q4: Do you have any comments or suggestions about the possible control on genetic testing services accessed via the Internet?

Scope of the review

26. Central to this discussion is an understanding of the definitions that we propose to use, including what we mean by the terms 'genetic test', 'genetic testing service' and 'direct to the public'. We have provided a glossary of terms used in this document, but we believe that the above concepts merit some discussion.

27. In the 1997 Code of Practice, the Advisory Committee on Genetic Testing adopted the following definition of a genetic test:

"A test to detect the presence or absence of, or change in, a particular gene or chromosome".

28. In HGC's previous report we adopted a very broad definition of personal genetic information⁷, which encompassed various methods of obtaining information about a person's genetic makeup. This included the direct analysis of DNA or of proteins and other body chemicals, the direct observation of certain physical features (such as eye colour) and of information about family history of disease.

6. Promoting Safe and Effective Genetic Testing in the United States. 1997. www.nhgri.nih.gov/elsi/tfgt_final/
⁷ "information about the genetic makeup of an identifiable person, whether derived directly from DNA (or other biochemical) testing methods or indirectly from any other source". Inside Information, 2002 p27.

29. Elsewhere in our work we have adopted a narrower definition of genetic testing which is based primarily on the direct analysis of DNA, chromosomes, proteins or other body chemicals. We have also noted the variety of definitions adopted by other committees in the UK and USA. In particular, there have been several distinctions between gene tests and genetic tests. The former term has been used to refer to direct analysis of DNA, whilst the latter tends to be used for a variety of diagnostic or imaging methods that provide information about the genetic nature of a condition or disease⁸.

30. There are some attractions in concentrating solely on gene tests, in that it is easy to define and categorise a test on DNA. However, certain features of body chemistry are wholly determined by our genes. To regulate tests on the genes, but not on the products of those genes, might be seen as arbitrary or inconsistent.

31. In considering a broader definition of genetic test we recognise that certain features of body chemistry used in a test procedure may be predominately but not wholly a reflection of the underlying genetic makeup of the individual. For example, a person's cholesterol level might have a strong genetic component but information about family history of heart disease would be needed to quantify this. The same might apply for other tests such as prostate specific antigen (PSA) or bone density tests. It would be difficult to know where to draw the line between 'genetic' and 'non-genetic' tests. It might be possible to say for any test that there is a numerical cut off point that distinguishes a genetic condition from other influences. But again, this would be difficult and probably arbitrary. Moreover, whether the test should be regulated at all could depend on the actual or potential result of the test. The actual test result would vary from person to person or possibly even from day to day in the same person. However, it might be possible to consider regulation on the implications of the potential test result.

Q5: Should any controls or regulations be confined to tests carried out on the genetic material itself, or should they encompass tests on other aspects of body chemistry that might be directly determined by information stored in the genetic material?

⁸ Zimmern, R.L. (1999) "Genetic Testing: a conceptual exploration", *Journal of Medical Ethics*, **25**:151-156.

32. Another important aspect of the review is to consider what constitutes a testing service that is offered direct to the public. The ACGT Code defines this as a service:

“...supplied by a supplier to the public, outside the context of conventional medical referral systems.”

33. We wish to consider whether this is still a realistic approach to adopt, or whether we should consider adopting a narrower definition focussing on the types of services that may lead to most concern. For example, we consider that this review should not include genetic testing services provided through professional private medical practice. But we have also considered the possibility of genetic testing services being offered by other health professionals who have received appropriate training. Such professionals might come from a variety of backgrounds, such as pharmacy or genetic counselling. If such groups are in future excluded from the definition of a direct to the public genetic testing services they will continue to be subject to existing legal controls and professional self-regulation.

<p>Q6: What are your views on which genetic testing services can be considered, now or in the future, to be defined as “direct to the public”.</p>

The types of genetic test available

34. The following are examples of genetic tests that are currently clinically available (see Glossary for an explanation of the terms used here):

- to determine carrier status for inherited recessive disorders;
- for inherited dominant and X-linked disorders;
- for some inherited adult onset disorders.

Such tests have either minor or major implications for the tested individual. There is no indication at present that companies would be interested in offering all of these tests directly to the public.

35. It is important to note that the nature of genetic tests vary considerably. In some cases they are relatively simple tests in one particular area of the gene (for example the number of repeats at the start of the gene associated with Huntington’s Disease). In others there are a variety of different mutations in a single gene, a few of which cause the majority of clinical

cases (for example the common mutations in the gene associated with cystic fibrosis). In other cases there are a large number of different mutations or complex rearrangements which require careful testing and/or interpretation of the results by comparison with affected relatives or other cases (for example, the variety of mutations associated with Duchenne muscular dystrophy or in the BRCA 1 and 2 genes associated with hereditary breast and ovarian cancer). The different technical aspects of such tests will largely determine the possibility of them being offered commercially at a reasonable price and with an acceptable level of accuracy.

36. There are potentially a large number of tests which analyse the small natural variations in the gene sequence of individuals. These may be used in tests:

- for genetic susceptibility to multi-factorial or acquired diseases;
- to determine the effectiveness or adverse effects of medicines (called pharmacogenetics), food, chemicals and other environmental factors;
- to provide diagnostic, prognostic and treatment information about diseases such as cancer.

37. There are several other types of gene test that rely on the analysis of repeated sections of DNA outside of the recognised genes (the so-called junk DNA) in order to make deductions about relatedness or ancestry. These may be used in tests:

- to determine parentage;
- to determine genealogical origins;
- for the detection or investigation of crime.

38. It is also possible that in the future genetic tests might become available purporting to identify genetic characteristics associated with behavioural characteristics (for example, aggression or proneness to addictive behaviour), specific learning difficulties (such as dyslexia) or for intelligence. The genetic basis of behavioural traits is a particularly controversial matter.

39. The technology of genetic tests is undergoing rapid change. Existing gene tests tend to rely on the extraction, amplification and detection of specific parts of the genetic material using

a series of complex chemical and enzyme treatments. Although these methods can be automated to a large degree, they still require complex and sophisticated material and equipment. However, we must consider the possible future situation where gene tests may be conducted using ‘solid state’ methods that owe much to modern electronics. This may allow small and relatively cheap devices that can rapidly test samples of blood or cheek cells. Such tests may also be able to test for multiple genes or polymorphisms simultaneously, so-called gene chips. Increasingly, genetic tests might be done via “point of care” devices.

Q7: What are the likely developments in the genetic knowledge and technology that might impact on the number and type of genetic tests offered direct to the public?

Should genetic tests be specially regulated?

40. A wide variety of medical tests are now offered directly to the public. These include cholesterol tests, tests for osteoporosis, for ulcers (*Helicobacter pylori*), sperm count tests, and PSA tests for the detection of prostate cancer. These tests are not generally considered genetic tests although they may reveal a condition which is affected by genetic characteristics. For example, a high level of cholesterol may be recorded because the person tested has familial hypercholesterolemia. The cholesterol test therefore becomes a test capable of revealing genetic characteristics although it does not involve directly looking for the presence of a particular gene. As the genetic basis of more common diseases becomes better understood, then the boundary between DNA tests and other tests will become less clear.

41. The easy availability of over-the-counter medical tests has implications for genetic testing. If the former tests are generally available, is it appropriate to restrict the direct availability of genetic tests? This would only be justifiable if genetic tests were of a fundamentally different nature from other medical tests.

42. Genetic test results are personal genetic information. In our recent report *Inside Information*, we identified a number of reasons why genetic information might be considered to merit special treatment. These included the implications that such information has for other people, the fact that it can have predictive value, and the fact that it can be easily obtained from the analysis of small samples. Some of these attributes, however, are present in other forms of medical information, and any case for affording special treatment to genetic

information would depend on factors such as the sensitivity of the information and the use which might be made of it. In accordance with this view, any case for controlling the availability of over the counter tests will depend on the potential impact of the tests rather than on the fact that the tests are genetic in nature.

Q8: What distinction, if any, should be made between genetic tests and other medical or health-related tests which might be offered direct to the public?

Q9: If testing for non-genetic conditions is available direct to the public with few restrictions, then is there a case for restricting the availability of testing for genetic conditions?

Q10: What distinction, if any, should be made between genetic tests intended for:

- predictive health purposes;**
- diagnostic purposes;**
- life-style or other purposes which have a limited impact on someone's life?**

The right to personal genetic information

43. Any system of regulation of direct to the public genetic testing would have to be based on the notion that the state is entitled to control the individual's access to genetic information about himself or herself. If the State is financing testing, then it is entitled to make a reasonable choice between those conditions for which testing is available at public expense and those for which it is not. If the State is not financing testing, the only grounds upon which it could restrict the availability of testing would be that the test in question is potentially harmful to the client or to some other person. The client's right to information then becomes limited by the State's right to protect its citizens. This, of course, is a paternalistic position, but it is a form of paternalism which society is prepared to exercise in some areas where the individual might be vulnerable and might be subject to exploitation or improper pressure.

44. In the case of genetic testing, the possible harm may be the psychological trauma of being given information that the recipient is not prepared for or which is not properly interpreted. There may also be concerns about exploitation of vulnerable or anxious persons by offering them tests which do not provide useful information and which may merely increase needless anxiety. Finally, society as a whole may be harmed by negative perceptions that may

accompany the trivialisation of some types of genetic testing, the effect of which may be to reduce confidence in genetic testing offered in a clinical context. We can see many parallels here with the separate debate about the provision of HIV testing.

45. There is a further consideration regarding the indirect effects on the NHS of widespread genetic testing. Clients who are concerned about the results of a genetic test will probably turn to their GP for advice and follow-up. This in turn may result in referral to a specialist service for confirmatory testing and possibly for counselling about the risk to relatives and to future generations. Whilst this may be a good thing in some cases, it does raise the possibility of an increase in the need for GPs to sympathetically assess and manage the “worried well”.

46. These possible objections must all be viewed against a strong sense of public entitlement to information. Human rights claims are also relevant: the Human Rights Act 1998 protects a right to private life, and it is a concomitant of privacy that the individual is allowed access to the information needed to take decisions about his or her private life. We have noted that people are increasingly interested in taking charge of their own health and of making decisions about treatment and lifestyles. We have also noted the interest that many have in establishing their true biological parents, although we do not consider this in detail in this review.

47. Some also do not wish to make use of existing medical services despite the safeguards of medical confidentiality. Many people choose to use home pregnancy testing kits, or other private medical services, for convenience and confidentiality. There may increasingly be concerns about the possibility of sensitive information having to be disclosed to insurance companies or prospective employers.

Q11: If people are interested in finding out personal genetic information about themselves, then should they be entitled to obtain such information in whatever way they wish?

48. We have in our previous work considered the potential relevance of genetic information to other family members. There are some complex and conflicting issues relating to the sharing of information within families, and of the moral claims that may be made by

others. In clinical practice it is possible to explore these issues in depth during pre-test consultation and counselling. This may not be possible where information is provided in a written form as part of a direct genetic testing service. It is an important aspect that we consider later when discussing the options for future regulation of direct genetic testing services.

Basic elements of consumer protection

49. There appear to us to be certain basic duties towards consumers and defensible ethical approaches that should form part of any direct genetic testing service. These are largely set out in the 1997 ACGT Code of Practice and relate to the quality, accuracy and reliability of the tests and any information for the consumer. We highlight some of the following issues which have arisen during our earlier work or in connection with our consideration of recent genetic services supplied to the public.

Consent

50. It is widely accepted that the rights of the individual should be respected. We have previously set out the principle of consent – that private genetic information about a person should generally not be obtained, held or communicated without that person’s free and informed consent. In our recent report called “Inside Information” we have considered some of the details of consent, especially in relation to children who may have the capacity to consent for some forms of testing, and for adults without capacity. The existing Code of Practice recognises that some children have capacity to consent to tests, and covers the recognised test of “Gillick competence” in relation to who may be supplied a test. We have noted the specific legislation that now exists in Scotland, which would need to be considered in any new Code. But there is perhaps a wider issue about whether there should be restrictions or some form of discouragement against parents who wish to have their (young) children tested using directly available testing services.

<p>Q12: Should direct genetic testing services be limited to adults capable of giving informed consent?</p>

Confidentiality

51. The importance of maintaining the confidentiality of genetic information is also an important principle that we have considered in some depth. However, we have recognised the importance of balancing the duty of confidentiality against broader considerations. For example, at present GPs hold lifetime health records that follow the patient from cradle to grave. The results of genetic tests may have important implications for the client's healthcare in the future. The existing Code of Practice encourages arrangements for ensuring that GPs get a copy of test results for the patient record. Occasionally a patient at a NHS genetic testing clinic may wish to use a pseudonym or otherwise ensure that test results are not sent to their GP. This may be due to concerns that such information will be made available to employers or insurance companies, or because of other concerns about confidentiality.

Q13: With appropriate consent, should results from direct genetic testing be recorded in the GP record? Should this be encouraged within any future regulatory system, and if so, how might this be done?

Security and storage of samples and data

52. An important element of privacy and confidentiality is the eventual fate of test results and samples. The Code of Practice requires that samples are destroyed after 3 months and identifiable client data after 12 months. We are aware of some commercial organisations that store and test samples from volunteers – with fully informed consent - in order to provide valuable research materials for companies and organisations. The samples are anonymised and any genetic information does not appear to be passed to the individual. There is a possibility that some companies offering genetic testing services may wish to retain samples – anonymised or identifiable – in order to support research themselves or with partner companies. This would be covered by the existing requirements under the DPA for explicit consent before further processing, unless the samples or data are anonymised. It would also be subject to the existing requirements of contract law.

Q14: What are your views on the arrangements for retaining and using samples and data?

Quality, accuracy and reliability of tests

53. We discuss above the arrangements for accreditation of laboratories and possibly for the CE marking of *in vitro* diagnostic devices. A further important element of any genetic test offered direct to the public is the assessment of the accuracy of the test, its scope and limitations. The existing Code of Practice requires such information to be made available to the client in a form that they can understand. It also includes a voluntary notification scheme to enable independent verification of the supporting data. In other countries there are similar arrangements to allow independent verification of the accuracy and reliability of tests. However, these are usually intended to support the use of such tests in clinical practice or major screening programmes. We must also consider alternative scenarios where a test is offered against a background of a less formal scientific evidence-base, but does not have significant health or other implications for that individual. We might also need to consider the evidence of efficacy associated with any recommended interventions, such as preventative dietary advice or complementary therapy.

Q15: Do you consider that there should be some independent process to review the accuracy and reliability of some or all types of genetic tests that are available direct to the public? If so, how might this be achieved?

Q16: Should the value of any pharmaceutical or life-style intervention that is linked to a genetic test be considered as part of any accreditation process? How would this relate to established mechanisms for professional self-regulation?

Customer information and support

54. The Code of Practice sets out a number of requirements to ensure that comprehensive and understandable information is given to the client. In normal clinical practice a genetic test for a serious inherited condition is accompanied by extensive pre- and post- test counselling with an experienced genetic counsellor or clinical geneticist. However, this is not always necessary when genetic testing is done as part of normal diagnosis or treatment by other clinical professions. The Code requires that appropriate arrangements are made for the patient, or their GP, to have access to appropriate counselling at no additional charge. This may be a lengthy process, and one that might cause difficulties if it diverts the limited number of trained genetic counsellors in the NHS. It is also arguable that many of the genetic tests

that will be part of direct genetic testing services will have fewer implications for the client than other tests or medical procedures for which counselling is not the norm.

Q17: For which types of test should access to appropriate pre- and post- test counselling be a requirement? Who should undertake this?

Q18: What are your views on the need for more general consumer information or education about the issues raised by direct to the public genetic testing?

Possible future approaches to the oversight of direct genetic testing services

55. Above we have set out the existing protections and the issues that might be faced in future genetic testing services supplied directly to the public. We are keeping an open mind about whether such services should be subject to any regulation, and if so, what form this should take. In view of this, we outline below a range of possible options. These range from completely freely available over-the-counter testing at one extreme to a complete ban at the other.

Option 1. No specific regulation.

56. It would be possible to leave the issue to the market and to rely on professional and business codes of ethics as well as the broader framework of commercial and consumer legislation. Those who wished to provide genetic tests direct to the public would be able to do so. The public would be free to find out information about themselves, in the same way that they can for other types of medical or parentage testing which are not specifically regulated. Under such a system, the issue of quality would be determined by market forces, with efficient and reliable testers attracting more clients than those whose services are sub-standard. The existing general controls on the supply and advertising of goods and services would protect consumer's rights. An example of this might be the widespread availability of home pregnancy testing kits or services that offer to test hair samples for vitamin and mineral deficiency. However, this option also recognises that the NHS would continue to provide the most common route for individuals to access appropriate genetic testing and consultation.

Q19: Do you believe that the option of no specific regulation is desirable?

Option 2. Voluntary regulation.

57. Under this option the direct offer of a genetic test to the public would be governed by a voluntary code of practice, in addition to the existing general framework. This Code would cover issues such as the taking of consent and the giving of information about the test to the client before it is taken. Requirements as to standards, quality and the provision of counselling could also be set out. There would be no theoretical restriction on the type of the genetic tests that could be offered directly to the public. But in practice, tests with serious health implications would need to be offered with similar service requirements as those currently provided by the NHS.

Q20: If you believe that there should be some form of regulation, then is a voluntary code of practice the best way to achieve this? If not, how else could this be done?

Q21: If a new voluntary Code of Practice were to be introduced, what issues do you think it should cover?

58. A voluntary Code of Practice might be introduced by the Government or one of its advisory bodies (as in the case of the ACGT Code of Practice) or by an industry body (as in the case of the Association of British Insurers Code of Practice on Genetics and Insurance). The latter approach would be more consistent with the approach to consumer codes being adopted by the Office of Fair Trading or the work of the National Consumer Council on 'soft law'. In a voluntary system there would be no legal sanction attached to a failure to comply with the terms of the code of practice. In accordance with the OFT core criteria, reliance would have to be placed on the willingness of providers to accept the code and any monitoring and compliance measure and appropriate sanctions if they were found to be in breach.

Q22: What are your views on who should be responsible for introducing and administering any new Code of Practice?

Option 3. Voluntary regulation with restrictions on the type of tests that can be offered

59. It would be possible to reinforce a voluntary code of practice with some restriction on the type of tests that could be offered. Tests which could have a high impact on the client (for example a test for a serious late-onset genetic disorder such as Huntington’s Disease) would not be considered suitable for a direct genetic testing service. In essence, this option represents the current position in the UK. However, there might be scope to include some form of additional statutory controls introduced using existing broad powers such as the Health Professions Council or the Fair Trading Act.

60. Such a system could allow for the classification of tests or services into two clear categories: those that are approved for direct offer to the public and those that are not approved for this purpose. This second category would include those tests or testing services which require the interpretative skills of genetic counsellors or medical practitioners, and those which have a potentially very serious psychological impact for the client. The categorisation of tests or services might have to be undertaken by an authority or body which would be in a position to assess new tests proposed for the market. Alternatively, a Code could contain clear criteria as to which tests could not be offered directly to the public. The Code of Practice could be introduced in either of the ways suggested in Option 2.

Q23: Should any new voluntary code of practice stipulate that certain tests should not be offered direct to the public? If so, which type of tests should not be offered?

Q24: If certain tests were not to be offered direct to the public, what process or criteria should be used to identify such tests?

Option 4. Strict regulation

61. This option would apply a system of regulation similar to that which controls prescription-only drugs or the supply of HIV-testing kits. Under this option, for example, it might be an offence for a person other than a registered medical practitioner (or other authorised person) to offer a “controlled” genetic test. Not all genetic tests need be included in the regulatory system; the offence would only be committed by tests identified as having a particularly high potential impact on the client or ones that require extensive analysis and

interpretation. Other tests could be provided direct to the public in accordance with one of the above options (ie with or without the additional requirements of a voluntary code of practice). Any such system is likely to require new primary legislation, and this would delay its introduction. Such legislation would also need to consider the mechanisms for identifying the restricted type of tests – which might be similar to those considered under Option 3 above.

Q25: Do you think that a strict regulation system, of the sort described in Option 4 above would be desirable?

Concluding remarks

62. We have been asked to conduct this review as a matter of urgency and have therefore only had limited time to prepare a consultation document. Our intention is to begin a dialogue on this topic over the next few months and to conduct face-to-face discussions with interested groups and individuals during the autumn. If you wish to be considered as part of this second phase of information-gathering please contact the HGC Secretariat at the Department of Health.

63. If there are other aspects of the possible control or regulation of genetic testing services offered direct to the public which you feel are important please include them in your response to this consultation.

Glossary of some technical terms and abbreviations

ACGT	Advisory Committee on Genetic Testing
Adult-onset disorder	A genetic condition that shows symptoms only in later (adult) life.
Carrier	A person who has one copy of a mutated gene causing an inherited recessive disorder (see below). They are not affected but may pass the mutated gene onto their children.
Gene test	A test that directly analyses DNA or gene sequence
Genealogy test	Genetic tests used in the study of family history and descent.
Genetic counsellor	In the UK, a non-medical health professional providing genetic counselling in a clinical setting. Genetic counselling is a communication process between the counsellor and the individual or family which deals with the medical and other issues associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family.
Genetic test	A test to detect the presence or absence of, or change in, a particular gene or chromosome (including indirect tests for a product indicative of a specific gene change in a person)
Inherited dominant disorder	A disorder where, for the person to be affected, the mutated gene only has to be inherited from one parent. Dominant disorders include Huntington's Disease and familial hypercholesterolaemia.
Inherited recessive disorder	A disorder where, for a person to be affected, the mutated gene has to be inherited from both parents. Recessive disorders include cystic fibrosis and sickle cell disease.
Paternity test	A test which uses DNA analysis to determine whether a man is the biological father of a particular child.
Pharmacogenetics	The study of how people respond differently to drugs due to their genetic makeup, in terms of both how well the drug will work and what side effects the person might suffer.
Single nucleotide polymorphism (SNP)	A variation in a single base (letter) of the DNA sequence which is present in different individuals and which may be associated with variations in susceptibility to diseases or the way that the body treats certain chemicals.
X-linked disorder	A disorder caused by a mutated gene on the X-chromosome. X-linked disorders usually only affect males but can be transmitted through healthy female carriers. Examples are haemophilia and Duchenne muscular dystrophy.

Human Genetics Commission
Working Group on Genetic Testing Services supplied Direct to the Public

Terms of Reference

Aim

To review genetic testing services currently being offered to the UK public, to examine the framework for such tests and to enable HGC to make recommendations to Ministers by December 2002.

Terms of Reference

- To consider current genetic testing services provided direct to the public and available in the UK and the adequacy of present direct and indirect controls in the UK (including services offered via the Internet), EU and overseas;
- To take evidence and consider likely developments in genetic testing technology, public attitudes, the control of other comparable services in the UK and the control of testing services, including advice on such matters as lifestyle or suggested medical treatments;
- To prepare a consultation document and to consider other methods for obtaining the views of stakeholders and the public;
- To inform HGC's recommendations to Health & Science Ministers, particularly on the possible need for any changes to the advisory and regulatory framework and the provision of advice to consumers.

Membership

Philip Webb (Chair)

Elizabeth Anionwu

Ruth Evans

Hilary Harris

Sandy McCall Smith

Patrick Morrison

Hilary Newiss

John Sulston

Veronica van Heyningen

Geoff Watts.

In vitro Diagnostic Medical Devices Regulations.

The Medical Devices Agency (MDA) is the UK Competent Authority for all three medical devices Directives including the IVD Directive. It has the authority to act on behalf of the Secretary of State for Health to ensure that the requirements of the Directives are carried out. This will include ensuring that all IVDs meet the essential requirements of the *In Vitro* Diagnostic Medical Device Directive and in so doing, do not compromise the health and safety of patients and users.

The IVD Regulations form part of general consumer protection legislation, and ensure that IVDs that have a specific medical purpose meet relevant essential safety, quality and performance requirements before being placed on the market with a CE mark. The Regulations came into force on 7th June 2000 with a transition period until December 2003. During this time, manufacturers may choose on which basis to place their device on the market. The Regulations require that manufacturers of IVDs bearing the CE mark must be able to demonstrate that the device will achieve the manufacturer's claimed performance.

The IVD Directive will introduce for the first time legislative controls dealing specifically with the safety, quality and performance of *in vitro* diagnostic medical devices (IVDs). IVDs will then have to meet relevant essential requirements before being placed on the market with a CE mark. The IVD Directive requires that the device must achieve specific analytical performances claimed by the manufacturer.

The IVD Directive does not cover the medical value of the result. For example, the Directive covers how well a test can detect a gene marker, but not how that result should be used to determine subsequent medical decisions. The IVD Directive also does not cover IVDs that do not have a specified medical purpose.

The UK Regulations implementing the Directive came into force in the UK in June 2000. Manufacturers now have a transitional period lasting until 7 December 2003 in which to either comply with existing relevant national laws - if any - or to comply with the Regulations and CE mark their devices. Until the end of the transition period, there is currently no UK legislation that deals specifically with IVDs.

Article 1(2)b from Directive 98/79/EC on *in vitro* diagnostic medical devices defines an IVD as follows:

"in vitro diagnostic medical device' means any medical device which is a reagent, reagent product, calibrator, control material, kit, instrument, apparatus, equipment, or system, whether used alone or in combination, intended by the manufacturer to be used in vitro for the examination of specimens, including blood and tissue donations, derived from the human body, solely or principally for the purpose of providing information:

- *concerning a physiological or pathological state, or*
- *concerning a congenital abnormality, or*
- *to determine the safety and compatibility with potential recipients, or*
- *to monitor therapeutic measures."*