



Human
Genetics
Commission

Genes direct

Ensuring the effective oversight of
genetic tests supplied directly
to the public

A report by the
Human Genetics Commission

March 2003

You can get copies of the full report and the summary by writing to:

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We drew on a wide range of evidence during our review.
This evidence and the full and summary report are available
on our website www.hgc.gov.uk/genesdirect/.

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Foreword

In February 2002, we were asked by Ministers to conduct a review of genetic testing services supplied direct to the public. We began our review thinking that it would be a relatively specialised and narrow subject of most interest to professionals, industry and some consumer organisations. We were surprised at the level of interest shown from our initial consultation, through our public consultations and surveys and during the Human Genetics Commission's (HGC) public meetings where we have discussed the review.

Our review was conducted by a Working Group chaired by Philip Webb who has worked tirelessly with fellow Commissioners, the public and media to ensure that this subject received a thorough examination. I am particularly grateful to Philip, the Working Group and the HGC Secretariat for introducing a range of interesting and novel ways of consulting key stakeholders and the wider public. The lessons that we have learned about consultation and dialogue will be useful for our future work.

In a little over a year since we began our work we have noted the rapid changes to the regulatory framework for medical devices and medicines which we have tried to take account of. This is our final report in which we make recommendations to Ministers. But we do not believe that this is the final word on the subject. Increased knowledge of our genes and new technology mean that more and more genetic testing is feasible, and people are becoming more interested in factors that affect their health. Both of these things open up opportunities for 'high street' genetic testing.

We feel that there are serious issues to be addressed so that individuals get good advice and guidance in a way that delivers benefits to health without abuses or mis-selling of these exciting new technologies. We do not believe that there is a case for banning direct access to genetic tests, but as the title of our report suggests that there is a need for effective oversight. We have made some suggestions as to how these matters may be dealt with, but it is for others to implement these recommendations and deal with the detail.

Helena Kennedy QC,
Chair, Human Genetics Commission





Summary

The Human Genetics Commission was asked by Health and Science Ministers to ‘give priority to a more thorough review of the provision of genetic tests to the public’. We started our work in early 2002 with a review of existing genetic tests available in the UK and began our consultations.

HGC is not a formally constituted regulatory body and most of our conclusions and recommendations are intended as a framework to guide regulatory bodies (Government, professional and industry bodies). We do not set out to create precise recommendations to regulate direct genetic testing, indeed we believe that there is no ‘one size fits all’ option to the complex variety of tests available now or in the near future. Instead, the best way of protecting the public is through a combination of legal controls on the sale of tests and professional self-regulation of those who might supply tests.

We have defined direct genetic testing as any test to detect differences in DNA, genes or a chromosome that is not provided as part of a medical consultation (1.19). We consulted on different regulatory options for the future, ranging from doing nothing to an outright ban on supplying tests direct to the public. We found that there was overall support for regulating such testing, but there were differing views as to how to do this. Some wanted to prevent direct genetic testing altogether, but many felt that a mixture of statutory and voluntary controls was most appropriate for a fast-moving technology like this.

We accept that the right to obtain information about oneself is an important right and the State should not intervene unless there is a risk of harm, particularly to vulnerable people like children or the elderly. We identified two possible broad ‘harms’ from direct genetic testing (3.7):

- the impact on individuals of misinterpreted or erroneous predictive health information which overstates the role of genetics in developing common diseases. This may result in delays in seeking proper medical advice (or seeking unnecessary medical treatment) or making expensive and unproven dietary or lifestyle changes
- the possibility that people can get inappropriate genetic tests on children or other adults without proper consent.

There are currently very few commercial genetic testing services in the UK but others are likely to join the market. Some may solely supply tests via NHS or private doctors and are therefore outside the scope of this review. Some may, increasingly, offer direct genetic tests via a non-medical intermediary such as a pharmacist or complementary therapists (which we term ‘direct to the public’). A further sub-set of direct genetic tests are those offered ‘direct to the consumer’ such as home testing kits or tests done by collecting a sample at home and sending it to a lab for testing. We believe that widespread direct genetic testing will put extra pressure on GPs for advice before or after a test. There might be related burdens on NHS laboratories to repeat and confirm test results.



We suggest a framework which can guide those bodies that are responsible for regulation to make sure that companies only market high quality tests, with good customer support and that they do not seek to misuse the power of modern genetics as a marketing tool. Our main conclusions and recommendations are as follows.

We recommend stricter controls on direct genetic testing, but we do not believe that there should be statutory prohibition of some, or all, direct genetic tests (3.24). This should not mean that people face difficulty accessing appropriate genetic testing or health information about themselves.

We feel strongly that there should be a well-funded NHS genetics service supported by a genetically literate primary care work force, which can properly manage and allow access to new predictive genetic tests that are being developed (3.30). This could involve the NHS providing ready access to testing services provided by commercial testing laboratories. It would enable predictive genetic testing to be retained within a well-respected model of continuing healthcare.

In view of this, we conclude that most genetic tests that provide predictive health information should not be offered as direct genetic tests (3.32). We think that it is a helpful analogy to consider the restrictions on medicines. Medicines are often only available with a doctor's prescription. But some may be provided via pharmacists and others, if they are low risk, can be bought in any shop.

If a company wants to provide a direct genetic test then it should have to convince a regulator that the test is suitable and that anyone involved in providing the test has the right training and expertise to give good quality advice to the consumer.

We have concerns about predictive genetic tests that are done at home ('direct to the consumer'; paragraph 3.34). This is because of the problems of providing full information so that the implications of the test can be properly understood. There is also a danger that children may be tested without proper lawful consent on behalf of the child. We have recommended a new offence of the misuse of genetic information that we feel must be introduced before such testing is acceptable.

The Government is already making some big changes to the legal and regulatory framework that will have an effect on direct genetic testing. The following proposals are intended as a framework that can guide those bodies that may be responsible for regulation in this area (3.39):

- We conclude that the creation of the **Medicines and Healthcare Products Regulatory Agency** (MHRA) provides an excellent opportunity to develop an appropriate regulatory framework for direct genetic tests before they are placed on the market. The MHRA will oversee European legislation that controls some aspects of commercial genetic test kits and laboratories. It could also play a key role in promoting high quality direct genetic testing, for example by overseeing wider aspects such as scientific quality and clinical utility of genetic tests and the advice that is given to customers (3.40).



- We welcome the proposed arrangements for reviewing genetic tests which will be introduced by the **UK Genetic Testing Network** (UK GTN) of the Genetics Commissioning Advisory Group (GenCAG). We believe that this work may provide useful basis for the oversight of direct genetic tests (3.45).
- We also note a possible role for a new **Human Tissue Authority** that has been proposed as part of revised legislation on human tissue and organs. Some direct testing laboratories may need to be licensed by the new Authority (3.48).
- The **Office of Fair Trading** promotes stringent self-regulatory Codes of Practice which could ensure that companies put proper procedures in place to support direct genetic testing services. This could include details of how consent is authenticated, how information is provided, how securely and for how long they will hold personal data and samples (3.51).
- The controls on testing companies should be backed up by improved and consistent professional training and standards. Any health professional or complementary therapist involved in providing direct genetic testing should operate under standards as stringent as those for doctors, nurses and pharmacists to ensure that they have the best interest of the individual at heart and are knowledgeable about genetics. The new **Council for the Regulation of Health Care Professionals** may have a role in promoting the required standards of professional self-regulation for several groups of health professionals. Other bodies overseeing complementary and alternative health practitioners should aim to develop comparable standards (3.56).
- We share the widespread concerns about the advertising of direct genetic tests and believe that it should be discouraged. We believe that the **Advertising Standards Authority** and the **Office of Fair Trading** should emphasis the need for responsible and accurate advertising of such products (3.59).
- We think that **consumer education** about genetic testing will play an important role in minimising the potential harms that may follow from direct genetic tests. We would like to see a broader Government effort to inform the public about all forms of predictive genetic testing and about which tests may be suitable for them. We would like funding to be made available to bodies like the Human Genetics Commission, NHS Direct or other independent and trusted bodies to provide impartial advice about direct genetic tests in order to empower consumers to make appropriate choices (3.62).
- We have concluded that we cannot easily control genetic tests that are available overseas via the **Internet**. However, we want to promote high standards of regulation in the UK and to liaise with regulators in other countries to achieve effective and harmonised national and international controls (3.63).

We accept that these are potentially far-reaching recommendations in an area where the science, the industry and the regulatory bodies are still developing. We therefore intend to continue to monitor this area and propose hosting a workshop or conference in the future to consider progress in the light of the response to our report (3.64).



We have also indicated that we will look further at the arrangements covering paternity testing to see what, if any, changes might need to be made in light of our report and the responses that we have heard (3.65). One other area of concern is the potential development and marketing of genetic tests for behavioural traits. We will continue to monitor developments in this area (3.66).



Introduction

1. When HGC was established in December 1999 it assumed responsibility for the publications of the Advisory Committee on Genetic Testing (ACGT) and in particular, the *Code of Practice and Guidance on Human Genetic Testing Services supplied Direct to the Public*. The Code¹ was prepared in response to a number of services that aimed to provide a testing service to advise whether a person was a carrier of cystic fibrosis. Subsequently, however, a market for carrier-testing did not emerge and there were few notifications of new services under the Code of Practice.

2. During 2001 and early 2002, HGC discussed a new direct genetic testing service offered by Sciona Ltd. The service was for a series of genetic tests to provide advice and information on appropriate diet and lifestyle. It was to be sold directly to consumers via health shops, as well as offered via GPs or dieticians. Our consideration of the extensive voluntary notification made by Sciona identified some concerns about the nature of the tests and the evidence supporting some of the claimed health benefits of the service.

3. We quickly concluded that this form of testing service was not envisaged in 1997 when the ACGT Code was published. In addition, the quasi-regulatory role that HGC was being expected to perform was somewhat at odds with our role of consulting openly and widely and providing strategic advice to Ministers. Ultimately, the consideration of the specific Sciona service, and the mild public controversy it generated, highlighted the need for a wider review of direct genetic testing services. To some extent this had been anticipated by the Government. In February 2002, Health and Science Ministers asked HGC to conduct a review of the provision of genetic tests direct to the public. In accepting this task, we made our specific concerns known to Ministers in May 2002. They agreed that we should not complete the assessment of the Sciona service pending a more detailed review of the basis for the voluntary Code. Subsequently, Sciona announced a new service which it proposed marketing only via certain professional groups, including doctors, nurses, dieticians and nutritionists.

4. We were asked to complete our review by the end of 2002. The complexity of the matter and the appointment of new members to HGC made us decide to delay our report slightly so that the revised HGC could adopt it in early 2003.

How we conducted the review

5. An HGC Working Group (see Annex 1) considered the lessons learned since the 1997 Code. It began a consultation exercise to seek views on the future of direct genetic testing services and to compare these with current and future developments in home testing kits for other medical conditions. The Working Group considered in some detail the wider social and ethical implications associated with genetic testing. We used as a basis some of the conclusions and recommendations that resulted from our report on personal genetic information², *Inside Information: balancing interests in the use of personal genetic data*, which was published in May 2002.

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

² See <http://www.hgc.gov.uk/insideinformation/>



- 6.** One of the major issues we felt merited consideration was the balance that must be sought between respecting that individuals are entitled to health information about themselves and protecting vulnerable individuals from inappropriate testing. We considered what basis there might be for any State interference. One important consideration was felt to be the wider impact on NHS resources, for example on GPs' time or on NHS laboratories that may be required to conduct additional testing.
- 7.** In July 2002 we published over 600 copies of a consultation document and sought views from the HGC Consultative Panel of those affected by a genetic condition. We received over 65 written responses covering a range of individuals, professional, academic, industry and consumer groups and regulatory agencies. Full details of the responses that we received, and links to the evidence, are at Annex 2. In the following sections we draw from these consultation exercises to give a sense of the views expressed. We then draw our own conclusions and make recommendations for future oversight of direct genetic testing services.
- 8.** While the consultation was underway, we held a series of 'evidence-gathering' meetings with key groups (Box 1) and individuals. We are grateful to all those who took time to respond and for helping the Working Group to gain an understanding of the wider aspects of the regulatory climate and to hear first-hand the views of industry, professional and patient groups and consumers.
- 9.** We recognised that the market for direct genetic testing services will be largely driven by consumer demand, so we felt that it was important to seek views from consumer organisations and the wider public. As well as our written consultation document, we held a public meeting in Belfast, conducted a series of focus groups and an Internet-based survey via YouGov and used the Democs process. Throughout the review we were also very grateful to other organisations that conducted public meetings to discuss many of these issues and who kindly provided us with information and updates.
- 10.** Commercial ventures have, understandably, good reasons to promote their products to consumers. The UK, which subscribes to broadly universal healthcare via the NHS, has not to date experienced strong commercial pressures to introduce genetic testing services. There is a greater potential for this in countries without a universal healthcare system, for example the United States of America. The arrangement for oversight of genetic testing services in the USA. were covered in some detail in a report³ by the US Secretary's Advisory Committee on Genetic Testing (SACGT). This report admirably addresses many of the concerns that we have identified and we broadly agree with the conclusions that they have reached.
- 11.** We summarise the development of similar controls in Australia, Canada, Europe and Japan (Annex 4) and consider how these may relate to the experience in the UK, and particularly the ability to ensure appropriate standards for genetic testing services that are accessed via the Internet. It is encouraging that there appears to be a developing international consensus by some of the leading industrial nations on the need for oversight of genetic tests, the claims that are made and the manner in which they are advertised.

³ NIH Secretary's Advisory Committee on Genetic Testing (July 2000) *Enhancing the Oversight of Genetic Tests*

**■ Box 1:****■ Organisations that gave evidence to the Working Group**

Advertising Standards Agency	Human Genetics Alert
BioIndustry Association	Medical Devices Agency
British Association of Nutritional Therapy	Medicines Control Agency
British In Vitro Diagnostic Association	UK Genetic Testing Network
Consumers' Association	National Pharmaceutical Association
Genetic Interest Group	Royal Pharmaceutical Society of Great Britain
GeneWatch UK	Sciona Ltd
Great Smokies Diagnostic Laboratories	UK Office of Fair Trading



The review

Current and future trends in genetic testing

1.1 When the Advisory Committee on Genetic Testing last considered this matter it expected there to be a rapid increase in the availability of direct genetic testing services, particularly carrier testing for recessive genetic conditions like cystic fibrosis (CF). This has not been the case and we heard from Dr Paul Debenham about the experiences of University Diagnostics which was one of two companies offering CF carrier testing in 1997. The service was initially promoted via the Cystic Fibrosis Trust, but the Trust subsequently changed its policy in favour of antenatal diagnosis. The uptake of the testing service fell sharply as University Diagnostics took a decision not to advertise the service more widely, and the service was eventually withdrawn in the face of low demand.

1.2 One reason why our review was felt to be timely was the rapid increase in the understanding of individual genetic variations and the development of rapid techniques for analysing specific differences at the molecular level. There has also been considerable excitement about the advances in human genetics in the past few years, particularly in the light of the near completion of the Human Genome Project. A number of responses pointed out that there were several inter-related developments relevant to considering future trends in direct genetic testing services. These include:

- A better understanding of the genetic basis for susceptibility to, and progression of, common diseases.
- A greater understanding of the individual variations in genetic sequences at specific locations, called single nucleotide polymorphisms or SNPs.
- The development of pharmacogenetics and the approval of medicines targeted at subsets of patients with clinical conditions that can be defined by their genetic profile.
- Improved and miniaturised technology for the extraction, purification and processing of DNA samples. This might lead to high-throughput laboratories or simplified tests which allow relatively unskilled operators to extract, prepare and test DNA.
- Developments in the arrangements underpinning clinical decisions, for example clearer treatment and prescribing guidelines, and computerised expert systems.
- Public demand for genetic tests that predict future ill health against a background which encourages people to take more responsibility for their own health.



1.3 Many of these developments are a vital part of the development of genetics in clinical practice and are the subject of major investment by pharmaceutical and diagnostic companies. Some of the responses helpfully detailed such developments, many of which related to pharmacogenetic testing. But we heard from some commercial organisations that before certain types of genetic tests become widespread within the NHS there might be a growing commercial market to supply such tests privately. The challenge for us in this review was to identify where commercial pressures, coupled with public demand, might lead to such tests being offered direct to consumers. If such tests are available outside of the NHS, then one important consideration for our review is who should offer genetic tests? What information should be provided before a test? There may also be a need for independent mechanisms to check claims made by commercial bodies. In some cases any oversight of the validity of a test will need to consider services where there is less than definitive evidence of the clinical utility of a test.

Commercial genetic testing services

1.4 We sought views on the future development of commercial genetic testing kits or services during our consultation. We met with representatives from the relevant trade associations (Mr Crispin Kirkman of the BioIndustry Association – BIA – and Ms Doris-Ann Williams of the British In-vitro Diagnostics Association; BIVDA). We also met representatives of companies that have or are actively promoting their services in the UK (LGC; Sciona Ltd and Great Smokies Diagnostic Laboratories).

1.5 Overall the UK currently represents a relatively small market for *in vitro* diagnostic tests, with BIVDA estimating the total market to be about £380 million per year. The spending per person is approximately half that of the United States. But some areas are growing fast, particularly with the use of rapid manual or self-test kits (those that are intended to be used by health professionals ‘near patient’, or by individuals themselves).

1.6 Genetic testing is a small part of the overall diagnostics market, but is likely to expand rapidly. It is difficult to obtain precise figures for genetic testing. According to the report by the SACGT in 1996 there were approximately 200 laboratories providing 175,000 genetic tests for over 300 diseases or conditions in the USA. In the UK it was reported⁴ that over 43,000 samples were tested in 1998-99 for a range of serious genetic conditions. This figure does not include tests for some blood disorders and cytogenetic testing for some conditions like Down’s syndrome.

1.7 The BIA stated that several of its members are actively involved in developing genetic tests. They are principally bioinformatics companies carrying out studies on links between genetic markers and disease which may be sold as a platform on which other companies can develop products. Companies in the USA had prepared genetic tests and were waiting for commercialisation opportunities, probably over the next couple of years. Gene chips were also being developed by several international companies.

⁴ *Laboratory services for genetics* (August 2000) Report of an expert working group to NHS Executive and the Human Genetics Commission (see www.doh.gov.uk/genetics)



1.8 GeneWatch UK provided us with a very thorough update on some commercial developments in the USA and UK. Not all of these developments are seeking to market tests direct to the public, although that is not to say that they do not raise concerns among some groups. The commercial approach taken by Myriad Genetics in marketing the BRCA1/2 test for hereditary breast cancer via health professionals was mentioned by several. Some of these issues – particularly around patenting and licensing of tests – fall outside of the remit of this review. However, GeneWatch also expressed some concern about the rush to develop genetic tests for common diseases.

1.9 We met Dr Chris Martin, (Chief Executive Officer) and Mr Matthew Bowcock, (Chairman) of Sciona Ltd as part of our review. In October 2001 Sciona had notified HGC of a direct genetic testing service called ‘You and Your Genes’ that they were marketing via the Body Shop (Box 2). We discussed the broader lessons to emerge from the early experiences with the ‘You and Your Genes’ service. We noted that their survey found that the majority of people who bought the service felt that it was ethical and of value to them.

1.10 Sciona commented on the gradual change in attitude and the increasing interest that individuals had in their own health. Because of this, significant investment has been made in the broad area of genetic testing in North America and Japan. Sciona felt that increased consumer demand for genetic testing was unavoidable, at least internationally, and it had to be managed and regulated rather than resisted altogether.

1.11 We also met with Mr Brad Rachman from Great Smokies Diagnostic Laboratories. They have been carrying out clinical laboratory testing for primary care physicians since 1987, under the national accreditation requirements of the US Clinical Laboratory Improvement Act and the College of American Pathologists. GSDL had developed SNP panels for chronic clinical conditions such as osteoporosis and cardiovascular disease. For practical and ethical reasons GSDL took the decision to market such tests via health care professionals.

1.12 GSDL appeared to recognise the difficulty of analysing and interpreting SNP results for complex diseases. They have trained 1500 accredited health practitioners in genomics in accordance with American Medical Association arrangements for continuing professional development. We did hear some concerns about the style of presentation at these training seminars.

1.13 The only criteria for ordering a test is that the professional is accredited in their own field and is permitted by the laws of the state, province or country in which they practice to order clinical laboratory testing. GSDL has conducted several training sessions in the UK and the service is being distributed via at least two companies, which market diagnostic tests and dietary supplements to doctors and a variety of complementary and alternative health practitioners.

**■ Box 2:****■ HGC's review of the 'You and Your Genes' genetic testing service**

In 2001 the HGC Genetic Services Subgroup considered a voluntary notification from Sciona Ltd under the ACGT Code of Practice. HGC visited Sciona's premises in April 2002 and discussed the service at several meetings (see www.hgc.gov.uk). The Subgroup did not reach a final conclusion on whether the service complied with the Code because of the wider review and commercial decisions by Sciona.

The 'You and Your Genes' genetic testing service, marketed via the Body Shop and the Internet, aimed to provide tailored diet and lifestyle advice to consumers. It was based on genetic tests for natural variations in genes that are linked to the way that vitamins are absorbed or harmful components of the diet or environment are processed in the body. Most of the genes tested have long been known to be associated with the processing of substances by the liver and to be part of important pathways for detoxifying chemicals in the body. There has been considerable research in their role in metabolising medicines and in the development of cancer (related to individual variations in the metabolism of carcinogenic chemicals). The Subgroup acknowledged that the genes being tested did play an important role in nutrition and metabolism.

However, in this complex area of human genetics they felt that there was not yet sufficient understanding of the interactions between genetic, diet and life-style factors in determining future health. They were not convinced of the predictive value of any particular polymorphism nor of the proven value of such genetic tests as indicators for dietary change in order to reduce the risk of ill health. Indeed, HGC noted that several important scientific reviews of original published reports concluded that there was little or no direct health benefit associated with screening for variations in some of the common genes.

“the available data so far failed to show any consistent and strong interaction between genotype and chemical induced cancer. It was unlikely that the interactions studied to date (which mainly concerned genes responsible for the metabolism of chemicals) were of importance to public health.”

Committee on Carcinogenicity of Chemicals
in Food, Consumer Products and the
Environment. June 2002

“a systematic screening of individuals at risk seems to make sense only in situations of well defined carcinogen exposure”

Taninger *et al* (1999) Mutation Research
436, p227-261

Despite the above conclusions about the link between certain genetic test results and the individual risk of cancer, the Subgroup felt that any potential customer concerns were not adequately addressed in the supporting literature.

**■ Box 2:****■ HGC's review of the 'You and Your Genes' genetic testing service (continued)**

Overall, the Subgroup felt that the service probably provided sensible dietary or lifestyle advice but which was freely available elsewhere. However, the use of genetic terminology gave an air of scientific credibility when there was little or no detailed peer-reviewed evidence in support of the assumption that genetic tests could be initiators of dietary change in order to reduce risks. Finally, the Subgroup was concerned about the potential for false reassurance from some of the tests. A test result, for example, that someone is tolerant of alcohol may reinforce an unhealthy diet or lifestyle.

“If a person followed all of this to the letter, they would find little value in the Sciona service, other than in knowing about their genetic profile.”

Sciona Ltd.

‘Your and Your Genes’ customer information

“Among those with binary, deterministic, views of genetic susceptibility, learning of the absence of a mutation that confers increased susceptibility could well lead to a sense of invulnerability”

Professor Theresa Marteau
(evidence to HGC).

Public demand for direct genetic testing services

1.14 As well as the formal written consultation exercise, we commissioned several surveys to canvass the views of the general public (Box 3) and details of these exercises and links to the full reports are in Annex 3. The impression we formed is that the public does not generally appear enthusiastic about direct genetic testing services, but some felt that there would be advantages in speed, convenience and confidentiality. The focus groups suggested that because the NHS provides genetic tests free at the point of delivery, there was little need for direct genetic testing. Most participants in the focus groups would want their doctor's advice and therefore saw the NHS as an essential part of any genetic testing.

1.15 The YouGov survey showed a similar apparent lack of awareness of, or interest in, direct genetic testing. Over 60% of people said that they were unlikely or very unlikely to use 'home genetic testing'. But most people would consider genetic testing if it was offered by their doctor to help diagnose a disease (81%) or offered as part of a general health check-up (71%).

1.16 We have also noted, however, in our discussions that when home testing kits for pregnancy were introduced there may have been a similar low demand. The YouGov survey showed that 40% of women had bought a home test kit (of which pregnancy testing is probably the most common). We feel now that most women would use a home



pregnancy testing kit before they approached their GP for a consultation. A similar gradual build up of public demand for some or all potential direct genetic testing cannot therefore be ruled out at this stage.

■ Box 3:

■ Wider public consultations

Consultative Panel:

We launched our consultation document in July 2002 at a meeting of the Consultative Panel of people affected by genetic disorders, attended by 40 panel members and HGC.

Belfast meeting:

We held a short public meeting following our plenary meeting in Belfast in September 2002 and encouraged the audience to informally discuss the issues raised during our consultation and discussions.

Focus groups:

8 focus groups of 8 people were held in November 2002. The groups were set up to cover a range of ages to make sure we heard from younger and older people and those with younger and older children, and were held in Reading, Watford, Leeds and Birmingham.

YouGov Internet survey:

An Internet survey was carried out between 27 December 2002 and 5 January 2003 with a nationally representative sample of 2510 people.

Deliberative Meetings Organised by Citizens (Democs):

The New Economics Foundation (NEF) conducted 6 events to test a new model for public consultation called Democs which enables people to run their own focus group-style events. NEF organised six events between the 7th and 17th of January 2003, three in London and one each in Bristol, Birmingham and Edinburgh. 47 people attended the events, including 14 Consultative Panel Members.



What to regulate and why?

1.17 Our consultation sought views on the definition of genetic testing. We consulted on whether this term should be interpreted narrowly (i.e. just to tests on DNA) or more broadly. A broader definition would include tests that indirectly provided information about genes by detecting or measuring a gene product (such as a protein or other specific chemical in the body) that is associated with a genetic condition.

1.18 We received a number of interesting responses on this point, which we might loosely categorise as follows:

- **Narrow** – the definition should be confined to direct information about gene sequences from DNA or RNA tests or protein analysis that directly relates to gene sequence. Most of these responders felt that the public were concerned about DNA/gene tests and that oversight should recognise this no matter if it was somewhat illogical to single such tests out from other health testing services. Some felt that broadening the definition would make any oversight cumbersome and would inevitably delay matters.
- **Broad** – the definition should cover all direct and indirect tests that give information about genetic conditions. The rationale for this seemed to be that indirect testing could be as significant as direct DNA tests and that by its nature indirect testing may be more prone to variations in analysis or interpretation. There was also a linked view that limiting oversight to DNA tests would create a loophole that might encourage the unrestricted proliferation of indirect testing methods.
- **Purpose-specific** – several responses qualified their comments about a definition by considering the purpose of a test. Some felt that any oversight should draw a distinction between predicting susceptibility to complex diseases and predictive testing for monogenic conditions or carrier status. Others felt that the penetrance of a genetic condition was relevant, others that the use of tests for the diagnosis of serious conditions or where there could be serious consequences for the consumer if a test was erroneous or the result misinterpreted, were relevant factors.

1.19 We have considered these points and have decided to retain the definition adopted by the ACGT, but noting the additions that were introduced by the Genetics and Insurance Committee in their criteria for genetic tests used by insurance companies. Therefore for the purposes of our report we will define genetic tests as:

a test to detect the presence or absence of, or change in, a particular gene or chromosome, including an indirect test for a gene product or other specific metabolite that is primarily indicative of a specific genetic change

1.20 We conclude that this definition is simple and sufficiently broad to cover the majority of predictive and diagnostic tests that are likely to be considered as possible direct genetic tests. It also covers other activities such as DNA paternity testing, but these have not been addressed in our report.



1.21 We also note the similar, but more detailed and comprehensive, definition of a genetic test adopted by the SACGT. It made clear, however, that “*tests that are used primarily for other purposes, but that may contribute to diagnosis of a genetic disease ... would not be covered.*” In our previous report we also noted the US Bill on genetic discrimination (as an example of recent US legislation) which defined genetic tests as “*the analysis of human DNA, RNA, chromosomes, proteins and metabolites that detect genotypes, mutations or chromosomal changes*”.

Other types of health tests available direct to the public

1.22 We did not want to confine our consideration to genetic tests in the narrow sense or to adopt a ‘genetic exceptionalism’ towards such tests. We therefore sought views about what distinction, if any, should be made between genetic tests and other forms of health-related test kits or services.

1.23 Some who we consulted, such as the Royal College of General Practitioners, were concerned about the apparent lack of regulatory oversight of health-related tests for osteoporosis, prostate specific antigen (PSA) or sperm counts, given the possibility of false or misleading results. There was a feeling that there should be regulation (but not necessarily restriction) for all tests supplied direct to the public when:

- the safety of the consumer may be compromised as a consequence of acting on a misinterpreted or an erroneous test result; or
- the purpose of the test is to make a diagnosis of or to predict a serious disease.

1.24 While we were preparing this report, the Consumers’ Association published a study⁵ of some home-testing services for non-genetic conditions and expressed concerns about the accuracy and reproducibility of a range of tests for conditions like osteoporosis, diabetes, cholesterol levels and prostate cancer. The Consumers’ Association also published⁶ a report on the Sciona genetic testing service that included the views of a clinical geneticist, genetics researcher and a nutritionist. We took note of these reports in our discussions.

1.25 We also heard a range of views that were familiar from our previous work on personal genetic information. Several responses commented about the possibility of conducting DNA tests on small samples and without consent. Others felt that genetic tests could provide predictive information for individuals and their families. However, it was noted that other types of health-related test could equally provide sensitive and intrusive information to individuals and their family.

1.26 Several responses highlighted the differences between direct health testing for general medical conditions and that for the sort of direct genetic testing services that have been marketed to date. This was usefully summarised by GeneWatch and the

⁵ ‘Don’t try these at home’. *Health Which?* December 2002, p10

⁶ ‘Health Genes’, *Health Which?* December 2002, p17

Christian Medical Fellowship along the lines that molecular tests (i.e. those on DNA) should be distinguished from other medical tests because:

- The results of general medical tests usually have immediate importance with action needed in the near future (e.g. pregnancy, PSA, osteoporosis) whereas a positive genetic test may not require such action.
- The results of general medical tests are usually relatively straightforward to interpret. The relationship between genes and disease is very complex and often poorly understood so interpreting such test results needs specialised expertise.
- The results of general medical tests do not usually have a health impact on other family members. This is not the case for genetic tests.
- The interaction of genetic and environmental factors is complex and could more readily lead to misunderstandings than with general medical tests.

1.27 There was a general acknowledgement that the public has a special view of genetic tests and DNA information, irrespective of whether this was justified or not. Professor Theresa Marteau commented that actual evidence of whether the public treated genetic test results differently was confused. There have been some studies on whether positive (abnormal) or negative (normal) genetic test results engender a sense of fatalism or invulnerability, but the results are not clear and may not extend to self-selected tests such as those we are considering.

1.28 There is also the question of whether individuals have a binary, deterministic, view of genetic test results that might lead to false reassurance. The example given was if a 'lifestyle' test showed that a person's metabolism dealt effectively with alcohol this might lead to a sense of invulnerability and to excessive drinking. We saw evidence of this from the focus groups – there was a view that genetic disease was different from other diseases and that they were inevitably more serious and incurable.

What constitutes a 'direct to the public' genetic test?

1.29 A further aspect of the review was to consider what we meant by 'direct to the public'. In the consultation we highlighted the position adopted by ACGT, which is that direct to the public means a genetic testing service offered outside of the normal process of medical referral. We asked whether 'medical referral' might be broadened to encompass other groups of healthcare professionals such as pharmacists.

1.30 For the most part the responses agreed that direct to the public meant a service or a testing kit provided outside of a medical consultation or referral. There seemed to be recognition that most genetic testing should involve some element of medical consultation. This was expressed by many of the written responses and in the public survey work that we conducted. Over 80% of people in the YouGov survey would want

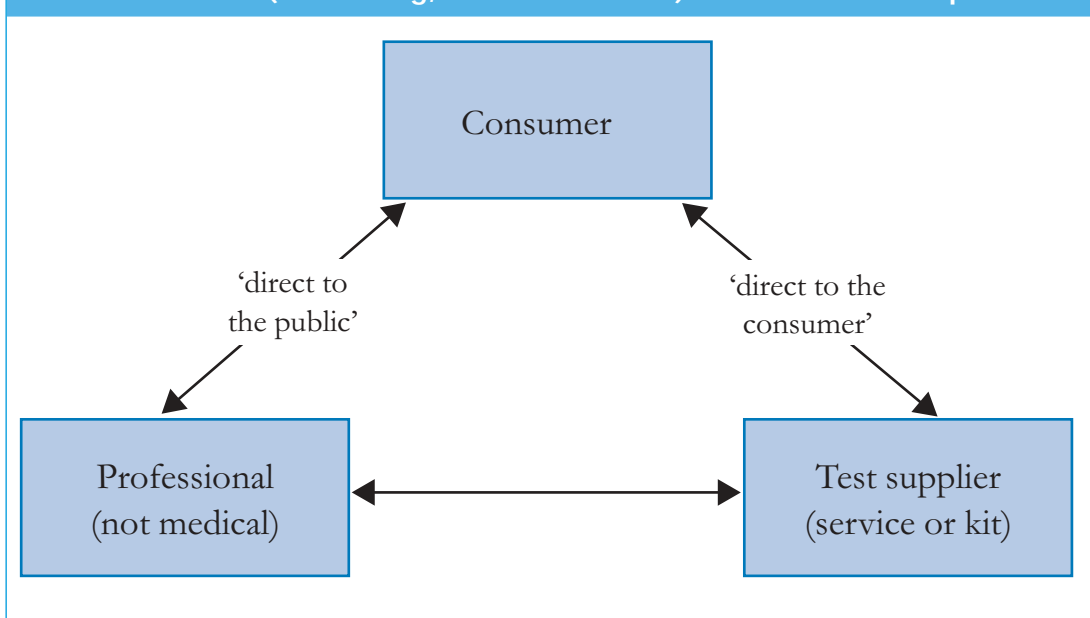


to consult their doctor for access to genetic tests for serious disorders or for carrier testing. About two-thirds would wish to involve their doctor for lifestyle tests and even for paternity tests.

1.31 There was, however, some recognition that the medical profession may also need to address the question of training in genetics. Even if they are not involved in the provision of direct genetic testing services, patients may approach their GPs for advice. Some also felt that there should be a regulatory mechanism to consider the accuracy and clinical utility of genetic tests provided within the NHS. It was remarked that GPs should then be able to advise patients about which tests are ‘accredited’ or ‘trustworthy’. They should also be able to direct patients to reliable services, and caution them about the advantages/disadvantages of any testing that is organised by individual patients themselves.

1.32 With this in mind, we concluded that we should focus our review on the future role of non-medical professionals in providing access to genetic testing services. In doing so, we reaffirmed the ACGT position that a direct genetic testing service means one offered outside of the process of normal medical referral. We therefore confine the majority of the recommendations to the role of healthcare professionals in providing ‘direct to the public’ genetic testing services (Figure 1) and to the possible concerns about a wider ‘direct to consumer’ model which may require further consideration.

Figure 1: Direct genetic testing services. The arrows represent both information flows (advertising, advice or results) and also DNA samples.



1.33 The BIA proposed a very narrow definition which usefully summarises what we term ‘direct to the consumer’ genetic testing service in which:

- A person buys the testing service from a retailer without any referral from a qualified medical practitioner.
- The kit or service allows the person to take their own biological samples, or samples are taken at the point of purchase by a non-medically qualified individual.



- The results of the test are provided directly to the consumer, either as a read-out from a measurement device or as a test report from a service centre, without referral through a medically-qualified practitioner and without counselling support.

1.34 Such ‘direct to the consumer’ genetic testing services (i.e. direct contact between a consumer and a test supplier, with the possible involvement of pre- and post-test counsellors) were anticipated as the major area of concern in the 1997 ACGT Code. We comment later about the desirability of such services.

The role of the health professions

1.35 Our consultation asked about the role of health professionals in the supply of genetic tests to the public. Healthcare professionals should be appropriately trained and able to provide impartial advice, in the best interests of the client, before and after testing. One point emerging in many responses was that high standards of professional conduct were necessary in the best interests of the ‘patient’ or ‘consumer’. This is particularly salient if provision is based on commercial interests. The Christian Medical Fellowship, for instance, stated that ‘profit must not be allowed to overrule the best interests of the patient’. One or two responses offered the suggestion that the role of healthcare professionals could be sub-divided into:

- tests that were offered in the context of professional advice and in which the healthcare professional was responsible for considering the best interests of the client, and
- tests where the healthcare professional was simply facilitating a transaction for a kit or self-testing mail-order service.

This distinction was felt by some to be an important factor in deciding whether a testing service should be considered to be ‘direct to the public’ or ‘direct to the consumer’.

1.36 The established professional bodies should continue to ensure the proper regulation of professional conduct and in setting and enforcing standards of professional practice. The Consumers’ Association and others felt that such bodies should cover the setting and maintaining of guidelines, standards for professional training, performance and conduct. They should also be responsible for training and accreditation of the healthcare professionals who would provide access to genetic testing service. Where professionals fail to meet set standards, such bodies would need to ensure that there are systems in place to protect patients from harm such as striking practitioners off professional registers.

1.37 Many responses commented on the main professional bodies for medical practitioners (the General Medical Council), nurses (the Nursing and Midwifery Council) and pharmacists (the Royal Pharmaceutical Society of Great Britain). We must also acknowledge the potential wider involvement of non-medical professional intermediaries and complementary and alternative health practitioners. We had detailed discussions with two groups relevant to direct genetic testing; pharmacists and nutritional therapists. These two groups demonstrated a marked difference in the level of training, accreditation and professional self-regulation.



1.38 Beyond the three main groups of health professions, many felt that there were important roles for a range of professionals. Several suggestions were made for groups that could be appropriately trained and accredited to use genetic information as part of their work. This would include professions that are regulated through statutory controls, but may include others with high standards of self-regulation. Examples mentioned included dietitians and physiotherapists. There may also be some laboratory staff who are members of the Association of Clinical Scientists. These groups are subject to regulation overseen by the Health Professions Council that began work in April 2002. There is also the Allied Health Professions Forum (AHPF) which represents the professions currently regulated by the Health Professions Council.

1.39 We believe that there may be an important role for the new over-arching body established by the NHS Reform and Health Care Professions Act 2002 to oversee aspects of professional self-regulation. When it begins work in April 2003, the new Council for the Regulation of Health Care Professionals will have oversight of the self-regulatory role of the main professional bodies such as the GMC, the Health Professions Council and the Royal Pharmaceutical Society of Great Britain.

1.40 The new Council will have the following functions that are relevant to our review:

- protecting the interests of the public and patients in the field of the regulation of health professionals;
- managing a framework for self-regulation, including oversight of regulators' rules and practices;
- requiring the regulatory bodies to conform to the principles of good regulation;
- promoting greater integration and co-ordination between the regulatory bodies and the sharing of good practice and information.

Pharmacists

1.41 We heard interesting comments about the possible benefits of groups such as pharmacists offering genetic testing services following self-referral, which was in part prompted by our separate consideration of pharmacogenetics. We therefore paid particular attention to the possible future role of pharmacists. We met with Ms Helen Darracot, the Head of Professional Standards at the Royal Pharmaceutical Society of Great Britain (RPSGB). The Society is the regulatory and professional body for pharmacists, including those working in High Street chemists, hospitals, industry and academia. It has similar functions to the General Medical Council (GMC). It also holds the registry of all pharmacy premises entitled to dispense prescription-only and pharmacy medicines. The Society had started work on the use of genetic information in drug prescribing. The Society is considering the requirements for quality assurance, training and patient counselling. Pharmacists would need to act responsibly, and this might be underpinned by amending the Society's Code of Ethics and Code of Practice. Current pharmacists may not have received training in genetics at undergraduate level. This was gradually changing and would be a key issue for pharmacists to cover as part of their continuing professional development.



1.42 The Society regulates its membership by imposing professional standards, and a person can be removed from the Register if necessary. There is a formal disciplinary procedure, which can also result in the company owning the pharmacy being disqualified from dispensing as well as the pharmacist being deregistered. The Society has an Inspectorate and visits every pharmacy once every 18-24 months. If there were concerns about a pharmacy or pharmacist, the first step would be to send an inspector to the pharmacy in question. If necessary, the inspector could advise the pharmacy not to offer a particular genetic testing service, but this is not currently enforceable.

1.43 Co-operation between pharmacists and GPs is very important. It was suggested that a pharmacist providing a genetic testing service should inform the local GPs. Such co-operation was linked to wider changes in the role of pharmacies in the prescribing process, with some successful pilots in community pharmacies which showed the benefits of links between GPs and pharmacists.

1.44 There is also the issue of adequate facilities – for example, a private consulting room in the pharmacy. We met with Ms Colette McCreedy of the National Pharmaceutical Association (NPA). The NPA represents most community pharmacies and provides services and representation about commercial and professional concerns. It tends to be concerned with the commercial aspects of running a pharmacist shop.

1.45 We heard that about 6 million people a day visit a pharmacy. These include a spectrum of the ill, the ‘worried well’ and hypochondriacs. They range from being highly educated people taking responsibility for their health to vulnerable people who could have undue faith in the power of genetics. Currently, pharmacists are paid by the NHS to dispense drugs, but they can also embark on commercial enterprises. Some pharmacists might be interested in providing genetic testing services because of their own interests or as part of their ongoing professional development, but it probably would not be a widespread activity.

1.46 The role of pharmacies is changing. We heard of consultations on the future skills mix in pharmacies that might allow pharmacists to spend more time consulting with patients and GPs. The NHS is setting up new services in deprived areas to provide care and counselling facilities. The recent changes to allow the morning-after pill to become available over the counter were also relevant because they pointed to the ability of pharmacies to provide facilities for counselling and dispensing in a sensitive and private manner, allowing a person to completely bypass normal GP consultations.

Alternative and complementary health practitioners

1.47 We formed the impression that there is a wider range of groups – which might be termed complementary therapists – who were interested potentially in providing genetic testing services. Some of the written response suggested that such groups might not be suitable to act as intermediaries in a genetic testing service. There were concerns about the role of voluntarily regulated complementary therapists, where the regulatory bodies that govern them have little power to protect patients.



1.48 Research by the Consumers' Association in 1999 showed that governing bodies have varying standards in the qualifications they require therapists to have before joining such that some accepted therapists after only a four-day course, while others insisted on two-years training or more. They also update their registers at different times, which could potentially leave practitioners on a list who are known to be guilty of poor practice. Some complaint procedures were also inadequate or non-existent.

1.49 We met with Sue McGinty and Amanda Whitewood from the British Association of Nutritional Therapy (BANT). Nutritional therapy takes the view that a person is what they eat and provides holistic advice on diet and lifestyle, often in close collaboration with NHS or private medical clinics. BANT publishes a Code of Ethics and Practice which details the role of nutritional therapists in relation to other health professionals and some of the legal restrictions. It also covers ethical issues such as conflicts of interest in relation to commercial diagnostic services or recommended supplements.

1.50 Presently there is no restriction on a person calling themselves a nutritional therapist. There are undergraduate and postgraduate degrees in nutritional therapy, but there is a wide variation in qualifications. Apart from a general requirement for nutritional therapists to be trained in a particular therapy, it did not appear to establish standards for such training or accredit its members and nor did there appear to be appropriate sanctions against those who breached the code of ethics and practice. This is changing with the development of National Occupational Standards for Nutritional Therapy and accreditation of qualifications via the Qualifications and Curriculum Authority.

1.51 Knowledge of genetics is certainly not a pre-requisite for nutritional therapists. However, there appeared to be a growing interest among nutritional therapists of the diet and lifestyle tests being developed and marketed by companies such as Sciona Ltd and Great Smokies Diagnostic Laboratories. We were concerned that BANT and its members appeared to have no mechanism for making judgements on the usefulness or otherwise of such tests. We formed the impression that they were content to rely largely on the claims made for testing services by the main commercial suppliers at training events and in company literature. BANT suggested that wider acceptance of peer-reviewed evidence surrounding the developments in 'nutrigenomics' would improve understanding of genetic tests for diet and lifestyle.

Is there a right to know genetic information about oneself?

1.52 In considering possible regulation of direct genetic testing we sought views on the extent to which individuals have a right to find out information about themselves and what role, if any the State may have in limiting access to such information.

1.53 The majority of responses to the consultation accepted that, in principle, individuals were entitled to receive high quality genetic information, via direct genetic testing, if they wished. Some felt that this was a natural extension of individual autonomy (or liberty), but that it was also important to pay attention to issues of consent. It was felt that individuals needed to know what tests entail and the

implications of test results for their future health. Inevitably, some respondents were concerned that commercial companies might not provide complete and impartial advice on the wider, perhaps negative, implications of genetic test results.

1.54 There was a clear view in some responses that it was wrong to speak of a ‘right’ to genetic information, as this implies that individual interest in personal genetic information outweighs family or societal concerns. Others pointed out that the information provided by direct testing would be more complex than simply detailing certain DNA sequences or SNPs. It would need to include information about the current state of knowledge about a person’s health and possible therapeutic or preventative measures that could be taken. This may be misleading or based on imperfect knowledge.

1.55 We also noted that there might be a role for State involvement if direct genetic testing resulted in increased burdens on NHS resources, such as increased calls on GP’s or clinical genetics professional’s time and on laboratory resources. Some drew parallels with private IVF treatment that may have led indirectly to an increased demand for neonatal specialist care for premature and multiple births.

1.56 The medical profession was clearly seen by the focus groups as the source of further information and treatment. Most participants said that they would seek advice from their GP unless test results from a direct service or kit were clearly negative. This was backed up by the YouGov survey.

1.57 There was also a perceptive comment that if an individual consulted their GP following a direct genetic testing service, the GP might feel obliged to request a re-test or other confirmatory testing in order to confirm or clarify the results. This would represent an additional call on NHS resources. If direct testing services are conducted properly, then this may result in timely and effective treatment or prevention options. However, if the direct testing service is of poor analytical or clinical validity, then it may lead directly to unnecessary burdens on NHS staff and facilities.

“If you did a test ... you’d go to the doctor... What’s the first thing they’d do? They’d send you for [another] test wouldn’t they?”

Focus group member

Consent and counselling

1.58 It is generally acknowledged that all genetic testing should only be done with informed consent. We did not detect any deviation from this underpinning principle, although there was some variation in the responses to two aspects of this. The first was whether direct genetic testing services should be allowed for young children, that is whether parents could consent on behalf of a child. The second related aspect was in what circumstances pre- and post- test counselling should be offered.

1.59 There was a general consensus about the difficulties of obtaining informed consent from or on behalf of young children. The need to consider whether the testing was in the best interest of the child – particularly for medical conditions – tended to suggest that direct testing services were not appropriate. The GIG response differed somewhat in suggesting that some types of test – such as testing for carrier status –



might be appropriate for young children. It is worth noting, however, that the 1997 Code stated that direct genetic testing services should not be supplied to those under the age of 16 and that the parents or persons with parental responsibility should consult their GP or medical practitioner for a referral to a specialist service.

1.60 We discussed children and genetic testing in *Inside Information*. The generally adopted principle governing the testing children for disease is that testing should only be undertaken in situations where it may have treatment implications in childhood. When an older child wishes to consider genetic testing the normal approach is to consider if they are competent to make a decision – which is generally known as Gillick competence. For the time being we see no compelling reasons to alter the sensible approach adopted in the 1997 Code, and recommend later that direct genetic testing kits or services should not normally be used in the case of young children. There may be some exceptions, such as the use of specific pharmacogenetic testing or of paternity and relationship testing. There are complex ethical issues involved in paternity and relationship testing and the legal situation is more complex. We intend to return to this topic in a further review.

1.61 Indeed our general concern about the potential harm of testing children makes us wary of any genetic testing kit or service that may be supplied direct to the consumer. Any genetic testing services that requires a sample to be collected at home or to be tested by the consumer at home runs the risk of samples being submitted for testing without proper consent. The difficulty associated with genetic tests that involved home sampling or testing was also identified by our focus groups.

“It was not clear ... how any company providing tests by mail order could verify that consent had been given or the age of the individual concerned” Focus group report

1.62 We have previously considered the potential for genetic testing to be done without adequate consent on behalf of the person being tested. This is a particular risk for young children and we have heard evidence of non-consensual paternity testing of children by men who want to check whether or not they are the father.

1.63 We recommended in our *Inside Information* report that a new offence⁷ should be created to deter individuals from taking samples from other individuals without consent. This should also be framed in such a way as to deter companies from offering services that may not have effective mechanisms in place to prevent non-consensual testing. We repeat our recommendation for a new offence to be introduced, and sincerely hope that the Government will respond positively.

1.64 Related to the provision of informed consent is the provision of adequate and appropriate information before and after tests. Most of the responses focused on the provision of extensive and non-directive pre- and post-test counselling which is considered good practice among the clinical genetics community. This was felt to be necessary for genetic tests that are predictive of serious conditions. Some felt that this would include predictive testing for single gene disorders and those that were predictive of a risk of cancer or other severe conditions.

⁷ “We recommend that consideration be given to the creation of a criminal offence of the non-consensual or deceitful testing and/or analysis of personal genetic information for non-medical purposes” HGC: *Inside Information*, p62. May 2002



1.65 Whilst supporting the need for counselling, the Genetic Interest Group (GIG) felt that there were situations where individuals may be sufficiently informed about their family history to not need extensive, and resource-intensive, counselling and support *“individuals should not be compelled to take part in such discussions; there are some well-informed individuals who may wish to avail themselves of a DNA test and nothing more, and they should be allowed to do so”*. Some would prefer to be able to access genetic testing services directly in order to obtain test results sooner or to maintain their own privacy and that of their family. However, we also note that clinical genetics centres are well able to adapt their service and even offer anonymous testing if so desired.

1.66 There was some variation in views about who should provide any counselling. Many commented that GPs and genetic counsellors were best qualified to provide this, but many also noted the impact this would have for NHS resources. Another view was that appropriately trained non-medical health professionals, such as pharmacists or dieticians, could provide this.

1.67 Some argued that for lifestyle and other testing for less serious conditions it might be appropriate to rely on written information, perhaps supported by telephone helplines. Against that, Human Genetics Alert (HGA) argued that carrier testing or pharmacogenetic testing provided potentially significant information and therefore may merit full counselling. This is not a view that we supported when we considered this in Inside Information.

1.68 We summarise later the ‘essential requirements’ that manufacturers are required by EU legislation to supply with self-testing kits. This includes the need for:

- Presentation of results in a way that is readily understood by the lay person... advice to the user on action to be taken if there is a positive, negative or indeterminate result and on the possibility of false positive or false negative result.
- A statement clearly directing that the user should not take any decision of medical relevance without first consulting his or her medical practitioner.
- Information about when a self-testing device is used for monitoring an existing disease the patient should only adapt the treatment if they have received the appropriate training to do so.

1.69 There is no statutory requirement to include such consumer-friendly information along with direct genetic testing services. However, the general principles outlined in the directive are sensible and we feel that this level of detailed and sensible information should be supplied as a minimum by those wishing to provide genetic testing services.



Information and advertising

1.70 There was a general view expressed that there should be some restrictions on advertising direct genetic testing services. The most often expressed concerns were that advertisements for services that were not scientifically valid could be misleading and result in unnecessary expense or worry. There were also particular concerns if, as a result of advertised services, consumers delayed proper medical advice or changed their diet, took dietary supplements or otherwise made changes to their lifestyle.

1.71 A large majority of the responses felt that there was a case for stricter, perhaps statutory, controls on advertising genetic testing services. This was perhaps summed up by the Consumers' Association response that "... *the situation is ripe for exploitation given the current level of scientific uncertainty, gaps in consumer protection and low levels of consumer education. For the foreseeable future we think that adverts for direct genetic testing services should be banned.*"

1.72 The marketing and advertising of direct genetic testing services was of concern to many in the wider public consultations. Advertising did not seem to be particularly likely to encourage people to use a genetic testing service. In our YouGov survey, 15% said that they would be more likely to take a test after having seen or heard an advertisement, versus 81% if recommended by their doctor or 49% via family and friends. The focus groups were also sceptical about a commercial market in genetic testing. Some felt that because a test would only be taken once, the real motive behind the test would be to sell other services or medication on the back of the test results. Some participants in the Democs process were concerned about the potential for simplistic and generalised promotion of the benefits of testing.

"... if the private sector was in charge of marketing they might simplify ... or generalise about the benefits .. whereas in reality the process of genetic testing can be much more complicated" Democs group

1.73 In our consultation document we summarised the current applicable controls on direct genetic testing. These include the Advertising Standards Authority (ASA) which administers the British Codes of Advertising and Sales Promotion⁸ for non-broadcast advertising. The Codes are enforced by the non-Governmental Committee of Advertising Practice, which is an independent committee whose members represent the advertising, sales promotion and media industries. The Codes stipulate that advertisements should be legal, decent, honest and truthful and prepared with a sense of responsibility to consumers and society.

1.74 We discussed the operation of the Codes with Mr Jim Barratt, former Head of Policy at the ASA. We noted that the Codes require that claims be substantiated and should not cause fear and distress. The burden of proof for any claims made in advertisements rests with the advertiser. In some cases, particularly with health products and therapies, the Codes makes additional restrictions to prevent advertising claims that may lead to misdiagnosis or prevent medical treatment.

⁸ Committee on Advertising Practice (April 2000) The British Codes of Advertising and Sales Promotion

1.75 We also noted that the Codes do not cover any advertisements directed only at medical, dental, veterinary and allied professions. The term ‘allied professions’ is open to interpretation by the ASA Council. However, we were reassured to note that it is not intended to be an open-ended loophole. Exemption from the Code is not given lightly, and consumer protection is a high priority.

1.76 Although a number of the consultation responses acknowledge the role of the ASA and the Codes, some felt that they should be strengthened and given additional support in relation to direct genetic testing services. This might include an additional section of the Codes with specific requirements, perhaps prepared in conjunction with HGC and the Department of Health.

1.77 During our review we became aware of the campaign by the Office of Fair Trading (OFT) about misleading advertisements and claims for ‘miracle-cure’ health, beauty and slimming products. The campaign draws attention to the Control of Misleading Advertisements Regulations 1988. These support the arrangements laid down in the CAP Codes, as well as those covering broadcast advertising controls. It provides a basis for the OFT or local Trading Standards officers to seek a court order to stop misleading advertisements.

1.78 Of particular relevance to our review is that the campaign aims to give consumers some common-sense tools to make informed choices about products. It emphasises, through amusing fictitious advertisements, that if a product or service ‘looks too good to be true, it usually is’. Some, for example, the Christian Medical Fellowship commented that *“there needs to be much greater education of the general public about the limitations and inaccuracies of genetic testing and prediction, and the unpredictable consequences of undergoing testing, before such tests become widely available”*.

1.79 In the UK there are strict regulations covering the advertising and promotion of prescription medicines (the Medicines (Advertising) Regulations 1994 – SI1994/1932). These prohibit the promotion to the public of prescription-only medicines. They also restrict the advertising of medicines and seek to ensure that claims are in accordance with the approved Summary of Product Characteristics. We heard from the Medicines Control Agency (MCA) about the detailed requirements of the regulations and the means of monitoring compliance with the regulations. The MCA is currently consulting on a slight relaxation of the controls on the advertising of certain medicines that are cleared for pharmacy sale or general sales.

1.80 Of interest to us was the availability, through the Internet and email, of foreign advertising that does not have to comply with UK standards. As an illustration, the websites of multi-national pharmaceutical companies must specify the target audience because of the differences in national laws on advertising medicines.

‘Direct to consumer’ advertising in other areas

1.81 Several responses, notably the Consumers’ Association (CA), drew parallels with the debate over direct-to-consumer (DTC) advertising of pharmaceuticals in the United States. According to CA, such advertising aims to increase sales, even if better options are available or the claims being made have not been fully substantiated. They argued



that advertising should not be confused with information. They also felt that there was a risk that advertising would instead provide misleading information that would ultimately lead to false consumer demand. There are also more subtle approaches to raising consumer or patient demand. The Nuffield Council on Bioethics report on genetics and behaviour⁹ highlights these in relation to the ‘medicalisation’ of personality traits in order to encourage sales of the newer classes of drug such as Ritalin and Prozac.

1.82 We also considered the developments in the US aimed at raising awareness of commercial genetic testing services that can be accessed via a medical practitioner in the light of a recent paper¹⁰ summarising some experiences. The authors concluded that DTC advertisements could overstate the value of genetic testing for clinical care. They may also exaggerate consumer fears about risk and about the deterministic nature of genetics.

1.83 Some respondents to our consultation specifically endorsed the 1997 US National Institutes of Health task force recommendation that advertising or marketing of predictive genetic tests to the public should be discouraged. The SACGT similarly recommended that the Food and Drug Administration (FDA) and the Federal Trade Commission should enforce the regulations in the area of genetic test promotion and marketing.

1.84 Whilst we were finalising our report we also noted the clear statement by the European Group on Ethics in Science and New Technologies about advertising of genetic tests via the Internet. They concluded that the information in such advertisements is likely to be misleading and incomplete, particularly in relation to complex genetic diseases. They also concluded that the advertising of genetic tests tends to convert them into commodities and to give rise to demand for testing that may result in social and personal conflicts.

1.85 The need to empower individuals to make informed choices has always been a fundamental aspect of genetics, as with most areas of medicine. The challenge in terms of direct genetic testing is to provide such support in a complex and fast-moving area of science. From the responses to our consultation we broadly conclude that extending the current UK restrictions on the advertising of medicines is appropriate for at least some types of direct genetic testing service. We will return to this later in the context of future regulatory oversight.

Confidentiality

1.86 We sought views about confidentiality of results from direct genetic testing services, and in particular, about whether results should be included in the individual’s GP health record. Most responses felt that it was important that test results were placed on a health record. However, most felt that the final choice should rest with the individual. The advantages of this for the future care of the patient should be explained to the customer and they should be given a copy of the results to pass to their GP.

⁹ Nuffield Council on Bioethics (2002) *Genetics and human behaviour: the ethical context*

¹⁰ Gollust, S. E., Chandros Hull, S. and Wilfond, B. S. (2002) Limitations of direct-to-consumer advertising for clinical genetic testing. *Journal of the American Medical Association* **288** 1762



1.87 It appeared to us that one important reason why people would access direct genetic testing services was to ensure that the results were not present on their GP records and therefore not likely to be disclosed by the GP in preparing a health report for insurers or employers.

1.88 We also sought views on the arrangements for the retention of samples and data by the testing company. Almost universally the responses indicated that any arrangements should be made clear to the individual and retention or further use should not be done without consent. The view was that the 1997 Code of Practice was probably acceptable in requiring retention of samples for 3 months and of data for 12 months.

“If you do [a] test at home would a company have access to your DNA, with related issues about access to information?”
Democ participant

1.89 Some responses were concerned that results from genetic tests could be used by the testing company or its affiliates for direct-marketing of medicines, vitamins or dietary supplements to those that are deemed to be ‘genetically susceptible’. Again, this is a matter that should be made clear – in the normal manner of contracts – when the individual agrees to the testing service.

Discrimination and stigmatisation

1.90 The risk (or perceived risk) of genetic discrimination is highly relevant to our review. We heard several concerns about the potential for fears about discrimination by employers or insurers driving people to access genetic tests anonymously via the Internet or commercial sources. Several comments supported this view and felt that there needed to be separate legislation to prevent genetic discrimination. This is an area where we have previously recommended consideration of legislation and we await the Government’s response.

HIV testing

1.91 The concerns about stigmatisation and discrimination raise parallels with the early days of testing for Human Immunodeficiency Virus (HIV). These were, in part, prompted by the position then taken by the insurance industry. The development of HIV test kits meant that, in the early 1990’s, there were concerns that ‘cowboy’ operators might enter the market and provide a poor service in relation to pre- and/or post-test discussions and the test kits themselves. It was argued that without proper advice and support an individual with a possibly unreliable positive test might become severely depressed or even suicidal.

1.92 In 1992, the Government acted on these and broader concerns about the public health implications of people opting out of the well-established arrangements for controlling the spread of sexually-transmitted infections. They recognised the importance of post-test discussions with a trained health professional in both dealing with the impact and consequences of a positive HIV test and as a means of communicating important advice on the prevention of transmission in relation to



negative HIV test results. The HIV Testing Kits and Services Regulations 1992 made it an offence to sell, supply or advertise for sale or supply an HIV testing kit or a component part, to a member of the public. As far as we are aware, these regulations have been effective.

1.93 There have, however, been some significant changes with respect to HIV testing in the international context, specifically in the USA. In 1996, the Food and Drug Administration approved the first HIV home sample collection testing service. This followed about a decade of debate as to whether or not home test kits should be available. The key concerns with the direct to the public HIV test kits were the accuracy of results and the ways users would be notified of test results. There were also concerns about how to ensure confidentiality, and how those who tested positive would be given support and advice. Similar sorts of concerns have been raised in the current UK context by a leading HIV and AIDS charity, the Terrence Higgins Trust. This needs to be put in context, however, that according to the Trust, it is recognised that home testing kits and kits purchased over the internet do ‘offer greater opportunities for accessing HIV testing for some people’.

1.94 The impact of the introduction of HIV home sample collection testing services in the USA has been mixed. While the usage of kits has not been as high as anticipated, thus having minimal impact on the testing behaviours of high-risk individuals, there is some evidence to indicate they may be a useful source of testing for those who would not use other testing routes, and for the ‘worried well’, those for whom the risk of infection is low but are seeking reassurance.

1.95 In the UK context, there have been some changes to the treatment of people with an HIV positive status and to public attitudes about HIV and AIDS. One of these changes is legislative, and includes plans to ensure that HIV infection is counted as a disability for the purposes of the Disability Discrimination Act 1995, from the point of diagnosis. Having said this, however, the UK approach to ensuring that HIV testing remains under medical supervision is largely unchanged and the Department of Health has advised us that there are currently no plans to amend the 1992 Regulations.

Regulation of direct genetic testing in the UK

Possible regulatory options

2.1 We consulted on four regulatory options that appeared to us to span the range of possibilities. The four options were also included in the topic guide of the focus group consultations and in the background for the Democs events. They were not specifically included in the YouGov survey, but it is possible to infer some conclusions on this aspect from the results.

2.2 At one extreme we considered no specific regulation of direct genetic testing services (Option 1) or an extension of the current self-regulation via a Code of Practice overseen by trade or other bodies (Option 2). A stricter regulatory option (Option 3) would be to refine the current mixture of self-regulation by adding some additional statutory controls. Finally, the option of specific statutory restrictions on direct genetic testing services (Option 4), similar to those for HIV, was considered.

2.3 We received a number of very well considered responses to the written consultation that can only be summarised with difficulty. We attempt to do this, however, in Table 1. As in our previous reports, we wish to emphasise that we did not approach the consultation results as a simple exercise in selecting the most popular or most favoured options. For example, we felt that it was important to note some of the pragmatic comments from the focus groups, Democs and written responses about the advantages of flexible and quick non-statutory controls in an area that is developing quickly.

2.4 What was readily apparent from all of the responses was that the option of no specific regulation (**Option 1**) was not supported. **Option 2** – for voluntary self-regulation – was supported by approximately 11 responses that answered this question, particularly those from the industry. There were, however a number of important qualifying remarks to the effect that the industry itself should not have responsibility for administering the voluntary arrangements, instead they should be overseen by other regulatory bodies (Table 1).

2.5 The idea of a mixture of voluntary self-regulation and statutory controls (**Option 3**) attracted most support (about 24 of the written responses) and was also supported by, or acceptable to, most of the participants in the Democs events.

2.6 The final option of specific statutory restrictions (**Option 4**) was strongly supported by most consumer groups and by a total of 13 out of approximately 66 responses that covered this point specifically.



Table 1: Summary of responses to the 4 regulatory options in the consultation

<p>Option 1: No specific regulation</p>	<p>There was no discernible support for this approach.</p> <p>Most felt that there was sufficient scientific uncertainty to require that consumers were protected from unscrupulous companies. Industry respondents felt that the risk to public confidence in genetics required specific regulation.</p>
<p>Option 2: Voluntary regulation</p>	<p>Around half of the responses, and a majority from the industry groups, supported voluntary self-regulation. The key advantages highlighted were the flexibility to adapt to changes in a rapidly developing scientific and commercial background.</p> <p>It was noted that the Code would need to draw together existing legal and professional obligations, so would not exist in a vacuum.</p> <p>Consumer groups and others felt that a voluntary Code was inadequate, particularly because of concerns about enforcement and compliance monitoring.</p> <p>A variety of bodies were suggested to oversee a new voluntary Code. Some felt that HGC was the appropriate body, others that HGC's role should be limited to providing advice to Government.</p> <p>Several commented that the Government, specifically the Department of Health or its agencies should have overall responsibility. The planned Medicines and Healthcare Products Regulatory Agency or the Commission for Health Audit and Inspection were suggested.</p> <p>Several also suggested a role for groups like the BIA, BIVDA and/or the ABPI. Other bodies included the GMC, National Institute for Biological Standards and Control.</p> <p>Several comments emphasised the importance of a recognised 'kitemark' which would demonstrate those companies and/or tests that complied with any Code and had been approved by an independent body.</p>
<p>Option 3: Voluntary regulation with restrictions</p>	<p>This option was supported by a majority of responses and only opposed by groups such as GeneWatch and CA who are against voluntary regulation, even with restrictions.</p> <p>The question about whether certain tests should be restricted prompted a range of potential criteria including: the severity of the genetic condition, the impact of the test (psychological or in driving changes to diet, lifestyle or medical treatment) or the type of testing service (self-test kits versus mail-order services).</p>
<p>Option 4: Strict regulation</p>	<p>This option attracted support from consumer groups. Many considered statutory restrictions might be necessary for at least some types of tests, with others being eligible for sale direct to the public. Some felt some tests could only be provided by medical practitioners, others that broader definition of competent individuals could be acceptable.</p> <p>Others, for example GeneWatch and Human Genetics Alert felt that statutory regulation should extend to genetic tests provided by NHS or private medical practitioners. There should be a new statutory body ("Offgene") to regulate all aspects of genetic testing.</p>

Current UK regulatory controls on direct genetic testing services

2.7 We have noted the regulatory approach to genetic testing services in a number of other industrialised countries (Annex 4). We were particularly interested to consider the approach taken in European Union (EU) countries which are obliged to implement relevant EU legislation. Of particular relevance here are the Medical Diagnostic Devices Directive (93/42/EEC, as amended) and the In Vitro Diagnostic Devices Directive (98/79/EC) – the IVD directive. We held discussions with representatives from the Medical Devices Agency (MDA) which is responsible for implementing this in the UK.

■ Box 4:

■ Definitions in the medical devices regulations

The definition of an in-vitro diagnostic medical device in the IVD directive is:

“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.”*

A ‘medical device’ is defined by the parent Medical Devices directive (93/42/EEC, as amended) as:

“any instrument, apparatus, appliance, material or other article, whether used alone or in combination, including the software necessary for its proper application intended by the manufacturer to be used for human beings for the purpose of:

- diagnosis, prevention, monitoring, treatment or alleviation of disease,*
- diagnosis, monitoring, treatment, alleviation or compensation for an injury or handicap,*
- investigation, replacement or modification of the anatomy or of a physiological process,*
- control of conception,*

and which does not achieve its principal intended action in or on the human body by pharmacological, immunological or metabolic means, but which may be assisted in its function by such means.”

2.8 It became apparent that the IVD directive, and the corresponding UK regulations (The Medical Devices Regulations 2002 SI2002/618), are open to differing interpretations in relation to some of the potential direct genetic testing services. In particular, we encountered uncertainty about the precise interpretation of the scope of the regulations because the stated purpose of some genetic testing – namely to give ‘lifestyle’ advice based on genetic polymorphisms – may not be covered by the definitions in the regulations (see Box 4).



2.9 For our purposes the key point in this definition is whether the stated purpose of the manufacturer was that the device is to be used for one of the purposes in the sub-definition, for example, the diagnosis or prevention of a disease. We sought clarification of whether the ‘lifestyle’ services provided by commercial genetic testing companies such as Sciona Ltd were covered by the Medical Devices Regulations. It was interesting to note the different interpretations made in written and oral evidence by a variety of groups. We have taken note of the definitive interpretation from the MDA that lifestyle tests are within scope because it sought to measure a physiological state, namely the presence of SNPs in particular metabolic genes and hence the activity of certain metabolic pathways. We conclude that there should be a very broad interpretation of the definitions as they apply to genetic testing kits or services. Where a genetic test – including a commercial service – is intended to measure a specific polymorphism in order to guide decisions about diet, exercise or for the prescription of a medicinal product, it would seem to us that this should be considered to be an IVD for the purposes of regulation. If there is any doubt about borderline cases we would encourage the industry to agree to voluntarily comply with the IVD Regulations and submit to the CE marking process. This would be in keeping with the existing arrangements in the UK to allow manufacturers to voluntarily subject new genetic testing kits or service to one of the more stringent review processes under the Regulations.

2.10 We were informed that the ‘purpose’ element of the definition means, for example, that breathalysers used by the police or drug detection kit used by employers to check their employees are **not** considered to be medical devices and are therefore not covered by the directive and UK regulations. Similarly, the definitions would exclude a DNA test aimed to provide information about family relationships (such as paternity tests). There is potentially a grey area in relation to genetic testing for behavioural traits such as IQ that are within the normal range (i.e. that are not classed as a disease or disability). These may fall outside of the definition of an IVD. They may also be deemed not to be appropriate for provision via the NHS. We consider this later, but note that such testing may in future require special consideration.

Oversight of laboratory-based genetic testing

2.11 The Regulations clearly apply to manufactured kits sold for use by clinical laboratories (for example to test ABO blood groups) and to self-test kits (such as home pregnancy testing kits). The status of commercial testing laboratories is more complex. We concluded that if a commercial company uses ‘in-house’ or purchased equipment or reagents to test human clinical samples as part of a commercial service, then it **is** considered to fall within the scope of the Regulations. It is important to note that in this respect, it is in effect treated as the manufacturer of a ‘kit’ which is placed on the market (i.e. it does not sell a kit as such, but uses one as part of a commercial service).

2.12 This distinction is important, because the MDA stressed that the Medical Devices Regulations and parent directives only require that the IVD conform to the essential requirements, that is, it performs as stated and does not harm users. The Regulations were not intended to cover wider aspects of quality of the service, qualifications of staff or the marketing of the service. These are covered by international quality assurance standards or general consumer protection laws.

2.13 We feel that the status of some tests and of testing services under the IVD directive is potentially ambiguous. We encourage the MDA to clarify the interpretation of the UK regulations as they apply to genetic testing by commercial companies. Given the apparent concerns about genetic testing in other EU countries, we feel that the MDA (as Competent Authority for the IVD directive) should continue to promote common European guidelines on the interpretation of the regulatory framework.

Requirements of the IVD directive and UK regulations

2.14 The Medical Devices Regulations categorise commercial tests that are within scope into four categories. The categories, in increasing order of oversight, are:

- **General**

These should not be put into service or placed on the market until the manufacturer has made a self-declaration of conformity with the ‘essential requirements’ that apply to the product for its intended purpose.

- **Self-testing kits** (those that are intended to be used by a member of the general public in a home environment).

The manufacturer must submit details of the device for independent certification which will decide if it is suitable for that category or if it should be considered under the higher risk categories.

- **‘List B’ kits or services** (specific conditions that are listed in Annex II of the Directive) which include some ‘genetic tests’ for hereditary conditions such as phenylketonuria, for HLA tissue typing and for Down’s syndrome testing.

- **‘List A’ kits or services** (as specified in Annex II to the Directive) for determining ABO and Rhesus blood groups, some infectious diseases, notable HIV and Hepatitis.

For devices listed under List A or B of Annex II of the IVD directive there are progressively more stringent processes aimed at ensuring high quality of the manufacturing and batch approval process.

2.15 The IVD directive provides details of the ‘essential requirements’ that are intended to ensure that devices achieve the performance stated by the manufacturer for a particular purpose and that they do not compromise the health and safety of patients and users. However, the emphasis of the IVD regulations is clearly on safety, quality and accuracy of the tests. It does not, and is not intended to, consider directly the wider questions of scientific validity or clinical utility.

2.16 We have not considered in detail the criteria used to determine how IVDs are categorised, but the categorisation appears largely to be dependent on the risk associated with their use (or misinterpretation). We understand that the Directive has a mechanism to allow updating of the Annexes. The foreseeable developments in commercial testing services, especially the production of genetic testing arrays (DNA testing chips) will undoubtedly introduce a number of new IVDs that pose equivalent ‘risks’ to those currently found in List B.



2.17 We suggest that the MDA, as UK Competent Authority for the IVD Directive, should continue to seek to ensure that the European Commission works proactively to update the Annexes to IVD Directive in light of anticipated developments, rather than reactively in the light of potential problems with particular genetic tests.

Enforcement

2.18 The MDA is not responsible for approving medical devices under the Regulations, this duty is the responsibility of approved ‘Notified Bodies’ in the UK and other EU countries. For most IVDs the enforcement of the Medical Devices Regulations falls on the Secretary of State and is the responsibility of the MDA. MDA employs 12 people full-time and others part-time looking at compliance. The Secretary of State has power to restrict availability of medical devices for public health reasons. This may be achieved by separate secondary legislation or by Restriction Notices under the Medical Devices Regulations.

2.19 One important consideration for any suggestion of statutory controls is the impact of international measures aimed at creating a Single European Market and general removal of trade tariffs and barriers. Whilst there are often clearly specified arrangements for protecting public health, the OFT commented that there were important international trade issues involved in any consideration of national regulations.

2.20 The Medical Devices Regulations do not contain specific advertising controls and MDA has no statutory powers to regulate advertising of IVD test kits. However, if they become aware of misleading or inappropriate advertising or marketing they will hold discussions with the company concerned. They may refer the matter to the Advertising Standards Authority or where appropriate, the OFT who can take action where necessary.

Legal requirements for consent and storage of DNA or tissue

2.21 The 1997 ACGT Code of Practice contains detailed provisions relating to consent before samples are tested and on the subsequent secure storage of DNA material in order to ensure confidentiality. These requirements are still valid, but the legal basis underpinning them has changed and is still changing.

2.22 HGC responded in 2002 to the Department of Health and Welsh Assembly Government consultation report called *Human Bodies, Human Choices*. The consultation formed part of the major review of legislation on the use of human organs and tissue following the public inquiries into organ retention at hospitals in Alder Hey and Bristol. We continue to feel that there is a strong case for ensuring that future human tissue legislation includes at least some recognition of the particular problems associated with human genetic material. We note that the new legal framework should provide a statutory basis for regulating all aspects of obtaining, storage, use and disposal of all tissue and organs.



2.23 The human organs and tissue consultation sought views on a comprehensive regulatory mechanism for all uses of human tissue and organs. This might include an over-arching new Human Tissue Authority with powers to require registration or licensing of laboratories that hold human organs or tissue. We commented on the apparent omission of genetic testing services that are supplied direct to the public (including genetic testing services, DNA paternity testing or genealogical analysis). Most such services require the obtaining of a sample of buccal cells or blood samples and the storage of DNA obtained from these samples. We do not yet know about the final Government proposals for regulation of human organs and tissue. However we feel that such legislation should ensure that certain minimum standards are followed by organisations and companies that provide genetic testing services.

Scientific and clinical quality of genetic tests

2.24 We sought views on whether there should be a mechanism to consider the scientific and clinical value of a genetic test. We also wished to explore whether there should be independent assessment of any interventions (such as diet, supplements or therapy) linked to a direct genetic test.

2.25 We emphasise, however, that the voluntary ACGT Code of Practice attempted to establish an independent oversight of some aspects of scientific and clinical value. The Code and supporting guidance set out the details of the arrangements for internal and external quality assurance that addressed some elements of the analytical validity. The remaining points were addressed via the voluntary notification arrangements which required, among other things, the following information:

- Peer-reviewed evidence of the value of the proposed service, including population data;
- The relevant standards of the manufacture of the reagents used;
- The accreditation scheme and external quality assurance scheme and internal quality controls;
- Details of the customer information, and any information for GPs, as well additional interpretative information.

2.26 We formed a clear impression from the written responses to the consultation that there need to be some procedures to ensure that genetic tests supplied direct to the public meet certain minimum standards of scientific and clinical utility. The terms used here may be worth covering in some detail.

2.27 The USA SACGT report contains a very comprehensive analysis of four main criteria that should be considered before a genetic test is considered for service. They clearly defined the following terms which we feel essentially cover the main areas of concern:

- **Analytical validity:** how well the test performs in the laboratory, does it always detect a particular gene mutation and is it always negative when the gene mutation is absent



- **Clinical validity:** the accuracy with which the test predicts the presence or absence of a clinical condition or predisposition
- **Clinical utility:** the value of the test results for the individual in order to guide them to make choices about preventative or treatment strategies.

2.28 The August 2000 report on Laboratory Genetic Services adopted similar definitions for analytical validity, clinical validity and clinical utility. It recommended that the Department of Health establish a national committee charged with the assessment and evaluation of new genetic tests according to agreed criteria. The written responses from GeneWatch and the Consumers' Association clearly linked direct genetic testing services to the arrangements being introduced for genetic testing services in the NHS. The Consumers' Association commented that *"the NHS system should be extended – or a parallel system set up – to cover commercially available genetic tests"*.

2.29 The Department of Health's Genetics Commissioning Advisory Group (GenCAG) was formed in 2001 as a sub-group of the National Specialist Commissioning Advisory Group to ensure that appropriate services are available in the NHS. One of GenCAG's terms of reference is 'to agree national criteria for the assessment and evaluation of genetic tests and technologies.' A new sub-group of GenCAG – the UK Genetic Testing Network (UK GTN) has been set up to advise on the implementation of the UK genetic testing network.

2.30 The Genetic Testing Network aims to provide high quality and equitable services for patients and their families who require genetic advice, diagnosis and management. They should be able to access a range of expert advice and appropriate tests via local genetics centres that provide the clinical interface and act as a gateway to a co-ordinated network of laboratory services throughout the UK.

2.31 We met with Professor Peter Farndon who chairs the UK GTN Steering Group and is a member of GenCAG. We were interested in the planned arrangements to evaluate the effectiveness of new tests before they are adopted as mainstream NHS services. The UK GTN will:

- Identify laboratory services that meet defined criteria for participation in the network.
- Oversee transparent processes over choice, evaluation and prioritisation of NHS funded tests available through the network.
- Encourage quality of service delivery to patients of clinically appropriate tests of acceptable quality.

2.32 The aim is to provide guidance for NHS commissioners to decide which tests the local genetics laboratories will provide on behalf of the UK network and which tests will be sent to other genetics laboratories in the UK network. The UK GTN will be supported by the National Genetics Reference Laboratories that have recently been established in Manchester (NorthWest) and Salisbury (Wessex).

2.33 We were interested in the criteria that the UK GTN Steering Group will develop. These are to cover the type of test, its accuracy, what information it provided, and its clinical utility. Of relevance to our review was Professor Farndon's view that there might

be two groups of tests, those provided free at the point of delivery via the NHS, and those accessible from the clinician but requiring payment. The reasons for their not being offered ‘on the NHS’ may be related to their perceived ‘cost-benefit’ in clinical terms. In this sense, the UK GTN will be acting in a similar way to the National Institute for Clinical Excellence (NICE) but its decisions are not binding. The UK GTN can only act by voluntary participation of laboratories, clinicians and commissioners. Although the steering group will consider best evidence for effectiveness and utility and make its recommendations it cannot enforce the required commissioning decisions to guarantee implementation.

2.34 A genetic test may not merit approval under the UK GTN process for two quite different reasons. One is that the test is less accurate or more complex and expensive than alternative tests. The other reason is that the test may not be supported by clear unequivocal scientific or clinical evidence of its value. This might be because peer-reviewed scientific publications or clinical studies are ongoing. It might be because the test does not cover a generally accepted medical condition. Either of these scenarios may be relevant to tests that are supplied direct to the public.

Oversight of any prevention or treatment linked to a genetic test

2.35 To some extent, the level of scrutiny appropriate for a test may hinge upon whether the test could lead to the need to embark on significant preventative or treatment procedures. When we asked about the oversight of advice that was linked to a genetic test we got a mixed response. There were a range of views, some of which appeared to be based on the assumption that HGC may be suggesting that it would have a role in commenting on the use of dietary supplements or medicinal products.

“[Companies] don’t just want to sell me one test kit. They want to sell me the insulin to use every day of my life.”

Focus group member

2.36 We do not consider that HGC, or other bodies primarily concerned with human genetics, can adequately address the claims made for dietary, lifestyle or other therapeutic interventions. We noted in our discussions that there are a plethora of parallel bodies that are responsible for considering such matters. Under the arrangements that we suggest later, it should be for a company to demonstrate to a regulator that it is following accepted medical, dietary or other advice about a particular course of action. Where at all possible, any responsible company should clearly demonstrate that the advice is not at odds with the regulatory requirements of, or the advice provided by, bodies such as:

- MCA (MHRA) – for medicinal products, herbal or homeopathic remedies;
- the Food Standards Agency (and Health Departments) for dietary advice and vitamin requirements;
- the National Institute for Clinical Excellence (where appropriate);
- professional and scientific bodies (such as the Royal Colleges, professional bodies or national bodies for alternative and complementary therapies), and;
- consumer bodies, patient organisations and health or medical charities.



2.37 We therefore do not draw any clear conclusions about the independent scrutiny of the advice or ‘treatment’ linked to a genetic testing service. We do feel, however, that the nature of the intervention will form part of the ‘informational impact’ of a genetic test. This in turn will be relevant to decisions about how to regulate particular genetic testing services available direct to the public. We will therefore cover this further in making suggestions for future regulatory oversight.

Genetics and behaviour

2.38 During our consultation, the Nuffield Council on Bioethics published its report on genetics and behaviour. It clearly points to the dangers associated with stigmatisation and discrimination in employment or education. It also highlighted the fact that a genetic test for gene variants that influence behaviour might be seen as comparable to personality test or IQ tests. Some tests may be associated with interventions that are seen by the providers and customers as similar to vitamin supplements or cosmetic surgery. Individuals may therefore be placed in a position where they take tests or consider interventions entirely outside of the normal arrangements for medical advice and consultation.

2.39 We do not yet know how soon genetic tests may be developed for behavioural traits. However, we do share the concerns of the Nuffield Council on Bioethics. Their report clearly identifies the current uncertainties in the scientific basis of behavioural genetics. It also notes separate developments in the ‘medicalisation’ of personality traits where there appear to be strong commercial pressures associated with the marketing of medicines. In the UK these pressures may be controlled to some extent by the arrangement under the National Institute for Clinical Excellence (NICE) to issue clinical guidelines for new medicines and new indications.

“Great caution is needed when measuring tendencies towards things like violence, rather than specific disorders...”

Democ participant

2.40 We agree that genetic tests for behavioural traits should be subject to strict regulation. We consider later the possibility that such tests will fall outside of the definition of a medical device for purposes of the Medical Devices Regulations. We therefore will make separate recommendations about the need for careful monitoring of this area of genetics.

Conclusions and recommendations

Key principles

3.1 The provision of genetic tests direct to the public raises important issues of principle. In our report, *Inside Information*, we identified a number of concerns over genetic testing in general, and proposed ways in which these might be addressed. We acknowledged that a distinction could be made between tests which might have considerable psychological or social impact on the person tested, and those that would not have this impact. Our recommendations in that report were that greater caution would be required in relation to high impact tests. It seems right to apply a similar distinction in relation to genetic tests offered direct to the public.

3.2 *Inside Information* dealt with issues of consent, confidentiality and discrimination in relation to genetic testing. All of these issues must be borne in mind in the provision of genetic tests direct to the public. Genetic testing, however provided, requires the consent of the person tested, and this consent requires a reasonable amount of knowledge of the implications of the test undertaken. The information that the test elicits should generally be treated as confidential, and it is important that those providing testing services should observe their obligation not to disclose such information to others without the consent of the person tested.

3.3 We also expressed concern in *Inside Information* about the possibility of wrongful discrimination on genetic grounds. Genetic information obtained from a test provided directly has the same potential for abuse of this sort as has genetic information obtained through testing in a medical context. It is important that those who obtain genetic information about themselves should be aware of any implications that this may have in matters such as employment or insurance. We have made general recommendations as to how the problem of discrimination may be tackled. These concerns apply equally to information obtained through a directly supplied test.

3.4 In addition to these general issues, which affect all genetic testing, the direct provision of genetic tests gives rise to a particular issue of principle: does the State have the right to regulate access to this form of information? This is a profound question, and how it is answered dictates the whole nature of the response to the issue. If the State does not have this right, then there should be nothing to stop anyone from offering the public whatever form of genetic testing they desire. It is notable that we did not receive any significant representations of this nature; nor did HGC take this view. We therefore accept that some form of regulation is justifiable.

3.5 The main argument against regulation is that to prevent somebody from obtaining knowledge about themselves infringes the individual's right of autonomy. An important value in our society is that we have the right to make decisions about our personal lives and that this power to decide should be restricted only when our decisions



might adversely affect the interests of others. This value has been of great importance in the area of medical decisions, where recent decades have seen a clear shift away from paternalism to the self-determination of the individual. Decisions about health matters are now seen as being matters for the individual whose entitlement to know about his or her health is an important aspect of the right of self-determination. In the past, people were denied access to their own medical records and were not always informed about treatments proposed for them – this paternalistic approach is now firmly rejected by modern medicine. It could therefore be argued that a clear burden lies upon those seeking to prevent access to information to justify why access should be denied.

3.6 This recognition of individual self-determination has a human rights dimension. The European Convention on Human Rights, which is now given domestic force through the Human Rights Act 1998, recognises the right to private life in Article 8. This article might be interpreted as preventing unnecessary restrictions on the right to find out information about oneself in order to make informed decisions about private matters. It is important to stress, though, that the State is still entitled to take measures to protect people from the harmful consequences of such decisions. What is required is that there should be a defensible balance between the exercise of this State power and the individual's right. We do not believe that the regulation of direct genetic testing would be an unreasonable restriction of the individual's rights under Article 8 of the Convention. Such regulation, however, would need to be justified in terms of prevention of undue physical or psychological harm to those who might seek such testing.

3.7 Ideally, people should be able to make informed decisions on the basis of assessment of risks and benefits. In practice, the information they are given may be limited, or may be of such a complex nature that it is difficult to understand. In these circumstances, some level of protection against exploitation is warranted, particularly where health matters are concerned. This is the justification for consumer protection relating to the advertising and marketing of health-related products and is also a well-established and widely accepted practice in relation to the provision of medicines. The State does not allow the uncontrolled marketing of pharmaceutical products or the uncontrolled offer of other therapies. In the case of pharmaceutical products, there are very strict controls on what can be offered, the justification being prevention of physical harm. The fact that the consumer might be willing to purchase a product and may regard the purchase as an exercise of individual self-determination is not sufficient to outweigh the need to protect individuals from self-harm. In particular, the need to protect vulnerable persons from harm is a strong factor at work here.

3.8 We believe that the right to know information about oneself is an important right, but that the possibility of exploitation and the causing of harm, particularly to vulnerable or unduly anxious persons, justifies a measure of regulation. This does not mean that the right to know is completely over-riden, but that its exercise is subject to some degree of restriction. In our conclusions below, we have sought to embody this balance between protection and free access.

3.9 We detected significant support for these views during our wider public consultation. There is strong public support for mechanisms to protect vulnerable individuals from misleading claims based on the widely held perception of the



predictiveness of genetics. Some individuals and organisations who responded to our consultation argued that rapid advances in our knowledge of genetics, and the understandable public interest in these advances, could be misused as a powerful marketing tool by unscrupulous companies in support of misleading claims.

“People will interpret information differently. They may not understand all the information and just pick up bits and pieces.”

Focus group member

Genetic testing and complex diseases

3.10 In our consultation we sought to distinguish between genetic tests which require careful analysis of multiple genes from those that involve only one gene. Diseases with simple Mendelian inheritance patterns are generally caused by mutations in a single gene, but often mutations in many different regions of the gene sequence can cause the same or related diseases. For these cases the whole gene has to be analysed in detail to be sure to pinpoint the disease-causing change. More complex diseases like heart disease or high blood pressure are usually caused by multiple predisposing variants in several genes. However, very often the actual disease causing changes have not been identified. It is through statistical analysis of general variation in the population that the patterns which mark the mutant gene causing disease are now being identified by geneticists. For such diseases the pattern of sequence variation at several selected gene positions is analysed in order to attempt to predict the level of an individual’s disease risk.

3.11 There have been several major research collaborations involving the sequencing of genes from many individuals. It has been possible to detect a large number of instances where the sequence in an individual differs from the ‘consensus’ human genome sequence at a single DNA base position. These are called single nucleotide polymorphisms (SNPs, pronounced ‘snips’) and have been extensively studied as a means of understanding the function of genes and of linking genes to the different features or disease patterns in individuals. Some SNPs are highly predictive of a certain feature (or phenotype), where for example they result in a different form of a protein or no protein at all. Others are simply markers for as yet undiscovered gene variants, but they allow some estimate to be made of the likelihood that an individual has a certain genetic makeup which might lead to disease.

3.12 There are an enormous number of potential SNPs in the human genome – up to 10 million by some estimates – and this has been felt to limit the use of SNPs to map and predict disease. Developments in human genetics have brought an increased understanding of how genes are grouped and structured into larger blocks called haplotypes. We heard separately from Professor David Goldstein about the use of haplotype mapping to reduce the number of SNPs that are needed to characterise individuals for the purposes of genetic research. These techniques may also result in more sophisticated methods of screening an individual’s genome for possible predictive health purposes.

3.13 Several of those we consulted pointed out that genes are poor predictors of complex diseases. With the exception of the widely used examples of genetic conditions caused by a changes in a single gene, there is still considerable uncertainty about the role



of genes and the environment in the progression of common complex diseases like cancer, cardiovascular disease, dementias or arthritis (Box 5).

3.14 We also received written comments, on a confidential basis, which commented on two of the commercial companies who provide direct genetic testing services. These commented that the training and supporting material somewhat over-simplified some of the complexities of the genetics of common disease and may have overstated the evidence that supported the use of their service.

3.15 There have been many reports of associations between genes and disease but these are not always reproducible. Even in relatively simple cases such as the enzymes that detoxify chemicals in the liver there is much confusion about the utility of genetic markers for predicting response to medicines.

3.16 There is an assumption of certainty in science that is often unjustified and this is especially the case in the genetics of so-called complex diseases. The term ‘complex disease’ applies to most major afflictions of the western world and describes conditions that emerge as a result of the interaction of genes and environment. This encompasses heart disease, blood pressure, diabetes, mental illnesses and most cancers.

3.17 Taking heart disease as an example, the idea is that genes make a person more or less likely to have heart problems, and this can be seen where there is a strong family history of either early heart attacks or long-life. A family history of early heart attacks does not necessarily mean that because your father had a heart attack you will too. Each individual inherits only one of the two copies of each gene from each parent. The combination of grandmother’s and grandfather’s genes inherited is more or less random and only some of the gene combinations are associated with risk of early heart disease. This is why detailed testing, with more knowledge than we have at present, will be needed to make predictions about our susceptibility to complex diseases.

3.18 But there are other factors involved and these are termed ‘environmental’, meaning that they have nothing to do with your genes. The environment in this setting describes anything that happens to you from the moment of conception (and possibly before), throughout life and it can also increase or decrease risk. Again taking heart disease, obvious things like smoking, diet and exercise will affect the chances of having a heart attack and, for the vast majority of people, will have a much bigger impact than any inherited susceptibility.

3.19 Although we recognise thousands of genes and are able to demonstrate variation within them, it is generally not possible to know what these variations mean. To extend the heart example further, blood levels of an amino acid called homocysteine affect the risk of premature heart attacks and these levels can be dependent on variations in the gene which produces an enzyme called MTHFR and is involved in the metabolism of folic acid. It might be thought that decreased activity of this gene would confer increased susceptibility to heart attacks, and there are studies that show exactly this. However, other, bigger studies, show exactly the opposite result. Does this mean that the gene is important in some people but not in others, and how do we recognise those in which the test may be useful? Or does it mean that some of the studies were wrong and others are right, and which ones fall into which category?

**■ Box 5:****■ The interpretation of genetic tests for common conditions**

- Even in highly penetrant major genes such as the BRCA1 and BRCA2 gene which are linked to breast cancer, carrying a faulty gene may only confer a risk of developing a condition (such as breast or ovarian cancer) in around 30-60% of patients.
- In complex disorders, such as, for example, raised blood pressure, factors such as weight loss, anti-hypertensive drug treatment and lifestyle factors such as smoking and diet, are significantly more important than genetic associations. Alteration of these factors may reduce risk by 50-80% (upwards or downwards) whereas genetic association data may alter risk by a factor of only 1-5%.
- Most complex disorders by their nature, will involve several genetic pathways within the body and measurement of 1 or 2 SNP's or modifier genes will only give an indication of possible risk and most reported associations to date, need to be confirmed in bigger or further studies.
- Interpretation of such results is necessarily complex and open to debate and will change as more evidence accumulates. We advise caution in interpreting present studies as insufficient data exists for accurate prediction of risk for most disorders.

Future oversight of direct genetic testing

3.20 The developing scientific and clinical understanding of genetics illustrates why it is premature to consider genetic testing as part of the normal clinical care of most diseases. For the immediate future, it is likely that only a small number of genetic tests might be generally available for health screening purposes. In 5-10 years, however, this position may well change radically. It is anticipated that the costs of genetic testing will fall considerably, and as the market for genetic testing develops, economies of scale will lead to the development of high-throughput testing, either to screen multiple samples for a few conditions or to screen one sample for many different genetic markers. The clinical value of genetic testing will also radically increase, first in the diagnosis and classification of diseases. This may then be followed quickly by predictive testing in order to guide the prevention or treatment of disease or for more general 'well-being' screening purposes. Before these tests are introduced into the NHS, they must meet certain standards of accuracy, cost and clinical value.

3.21 There are currently very few commercial genetic testing services in the UK but others are likely to join the market. Some may solely supply tests via NHS or private doctors and are outside this review. Some may, increasingly, offer tests via a non-medical intermediary such as a pharmacist or complementary therapists (which we term 'direct to the public'). A further sub-set of direct tests are those offered 'direct to the consumer' such as home testing kits or tests done by collecting a sample at home and sending it to a lab for testing. We heard views that widespread advertising of commercial genetic testing services (including direct genetic tests) will put extra pressure on GPs for advice before or after a test. There might be related burdens on NHS laboratories to repeat and confirm test results or to investigate conditions further.



3.22 This inevitably raises the question of how to effectively regulate direct testing services relating to common gene variations that may have a greater or lesser predictive value. We have considered the wide range of relevant public and professional bodies that are relevant to some aspects of genetic testing services and given some consideration to the current range of applicable legislation, both in our initial consultation and in subsequent discussions. There are a number of general consumer protection laws that will apply to aspects of genetic testing services. There are also some specific laws such as the 1939 Cancer Act and the 1992 HIV regulations.

3.23 Although we detected support for our earlier Option 4 (statutory regulation) we are concerned that such legislation would be extremely difficult to define. To take the example of the 1992 HIV regulations, these were introduced using extremely specific powers in primary legislation – the Health and Medicines Act 1988. These powers cannot be used to deal with any other diagnostic tests, even for communicable diseases that raise similar concerns to HIV. Even if we were to recommend similar powers for a genetic condition, the primary legislation would need to be drawn much more widely than the 1988 Act. It could also take many years to complete the Parliamentary process, by which time the science and commercial market may have advanced considerably. We must also acknowledge that there is no evidence of any real public danger or nuisance resulting from the small number of tests and services currently being marketed. Even if this were to occur in future, there are mechanisms in current law to restrict the availability of some medical devices (and therefore of commercial genetic tests).

3.24 We recommend stricter controls on direct genetic testing, but we do not believe that there should be statutory prohibition of some, or all, direct genetic tests. For legal and pragmatic reasons, we prefer to focus our attention on achieving a composite of controls based on general and specific legislation, regulatory bodies of several different complexions and a well supported series of Codes or guidance to ensure that the industry maintains high technical and ethical standards. (To return to the options in our original consultation, our recommendations are therefore for a mixture of Options 3 and 4.) We suggest a framework which can guide those bodies that are responsible for regulation to make sure that companies only market high quality tests, with good customer support and that they do not seek to misuse the power of modern genetics as a marketing tool.

3.25 In our review we have drawn parallels between genetic tests and medicines. Although it is not a perfect analogy, we note that medicinal products are broadly categorised into prescription-only, pharmacy-only and general sale medicines. Medicines are also subject to strict controls on advertising and marketing in order to protect individuals from unfounded or misleading claims. We also note the less stringent arrangements made by regulatory bodies to include homeopathic remedies where the scientific evidence of efficacy does not always meet clinically accepted standards.

3.26 The analogy with pharmaceutical regulation may be challenged on the grounds that there is a difference between taking a drug and obtaining genetic information about oneself. This distinction relied, however, on treating psychological and physical harm as quite distinct. In fact, the two forms of harm are inter-related: psychological harm has physical consequences. The consequences of knowledge about a genetic condition may have a profound effect on the well-being of the person to whom the knowledge is imparted.



The role of the NHS

3.27 We found strong public support for the NHS and GPs in giving appropriate medical advice before and after test – providing continuity of care, appropriate treatment or referral, as well as keeping medical records to help manage the care of patients. In our view, most predictive genetic tests properly belong as part of a consultation with a registered medical practitioner, either at primary care level, or for complex and highly penetrant conditions, at consultant-led specialist clinics. We also consider that this should include established screening and testing procedures conducted under the overall supervision of a medical practitioner (for example the infant heel-prick (Guthrie) test conducted by midwives or nurses). In other words, and to extend our analogy, we conclude that there is support for considering most genetic tests as if they were ‘prescription-only’.

3.28 We consider that medical practitioners are the most appropriate professional group for a variety of reasons. Most genetic testing should continue to begin at the primary care level because GPs:

- are well-regulated with a highly developed ethical approach and with strong sanctions against wrong-doing;
- have experience of the patient’s needs within a broad healthcare context;
- undertake formal training programmes leading to professional accreditation and with mandatory revalidation;
- contribute to and access the patient’s lifetime healthcare record, and;
- have appropriate facilities for a private consultation.

3.29 The public also supports a major role for the NHS in providing genetic testing services and advice and treatment in the light of such tests. Our YouGov survey suggested that around two-thirds of people would like to discuss a test and to receive results face-to-face with their doctor. Only 7% would prefer to receive information from a doctor who works for the testing company. For genetic tests for serious disorders, over 80% would expect to involve a GP or NHS specialist. This contrasts with just 10% who would prefer to obtain a test through a private consultation with a health professional. We therefore sense support for the view that, at present, predictive genetic tests for health and other purposes properly belong as part of a consultation with a doctor.

3.30 **We feel strongly that there should be a well-funded NHS genetics service supported by a genetically literate primary care work force, which can properly manage and allow access to new predictive genetic tests that are being developed.** Where genetic tests are clinically proven they should be potentially available on the NHS. Genetic services – clinicians, counsellors and laboratory support – need to be properly resourced and artificial access barriers (such as local variations in commissioning of services) removed so that patients can easily access genetic tests which have demonstrable benefit for purposes of health or reproductive choice. We look forward to the Government Green Paper on Genetics for reassurance that this is to be the case.



3.31 We emphasise, however, that within this paradigm that we have no inherent difficulty with the provision of genetic tests by commercial companies. New commercial services could be accessed via private or NHS medical referral, provided that they are subjected to appropriate regulatory oversight. We note that there is a potential role here for the proposed Commission for Health Audit and Inspection (CHAI) which will take over from the National Care Standards Commission and regulate certain types of private medical practitioners and private walk-in medical centres.

Direct genetic testing

3.32 In view of the above, **we conclude that for the foreseeable future, most genetic test that provide predictive health information should not be offered as direct genetic tests.** We think that the *presumption* should be that a genetic test that is predictive of a medical condition is generally unsuitable for supply ‘direct to the public’ via a non-medical health professional or other intermediary and unsuitable for a kit or services supplied directly to the consumer.

3.33 The regulatory approach we favour is that if a company wants to provide a direct genetic test via such routes then it should have to convince a regulator that the test is sufficiently well validated and that anyone involved in providing the test has the right training and expertise to give good quality advice to the consumer. For example, we recognise that certain genetic tests to guide the prescribing of medicines might properly be provided via certain pharmacists. A company would need to convince a regulator that a particular service was eligible to be offered ‘over-the-counter’ via pharmacists. The pharmacist would have to meet necessary professional standards of competence in genetics and have suitable facilities. This is analogous to the approach for medicinal products to be classed as ‘pharmacy-only’. We comment further on this below in relation to professional self-regulation.

3.34 **We have concerns about predictive genetic tests that are done at home** (kits or services that are supplied ‘direct to the consumer’). There are two broad types:

- home-test kits where the sample is taken by the customer and the results appear ‘before their eyes’ at home; or
- home sample collection where the sample is taken by the customer and sent to a laboratory for analysis. The results will generally be sent by letter, email or via the telephone.

3.35 We do not consider that current DNA testing technology allows genetic testing to be offered as **home-testing kits**. There are undoubtedly some testing technologies that rely on surrogates for genetic markers, for example proteins or metabolites, but these may be prone to inaccuracies. We comment above on the adequacy of the current regulatory system for considering such tests.

3.36 We are more concerned about services that rely on the use of **home-sampling**, in which a sampling kit, such as a mouth swab, is sent to the customer and returned to the testing company by post. These could potentially lack sufficient consumer support in ensuring that the customer understands what they are consenting to. There may also be difficulties in ensuring that the customer understands the implications of the test results, with potential knock-on effects on GPs and other NHS resources.

3.37 Home-testing or -sampling also raises the possibility of non-consensual testing of children or other adults. We have previously recommended that such testing be made a criminal offence and we reiterate these concerns. We have noted that the proposed new law on human organs and tissues should introduce much clearer duties to ensure that human tissue is only collected, stored and used with proper, lawful consent. We have earlier recommended that these controls should extend to genetic testing of all types of human tissue. If so, it means that companies will have clear legal duties to ensure that any human tissue that they collect and test has proper consent from the correct individual. The net effect of this may be to reduce the availability of home sampling and mail order testing services in favour of some form of direct face-to-face contact. In our view this ideally would be with a health professional or other responsible individual who can take proper consent and ensure that the sample is from whom it purports to be from.

The wider regulatory framework

3.38 We have considered the current UK regulations applying to genetic tests before they are marketed, as well as the plans for procedures to consider genetic testing services within the NHS. We have also paid careful attention to the report of the USA's Secretary's Advisory Committee on Genetic Testing and the response by the Food and Drug Administration (Annex 4). While we were conducting this review we became aware of a number of new regulatory bodies that appear to be relevant to the control of direct genetic testing (see figure 2). The Government is merging the regulatory agencies that deal with medicines and medical devices. It will oversee EU legislation to regulate some aspects of the sale of genetic tests. Other new or proposed bodies will oversee standards among health professionals as well as the use of human organs and tissues. We have also been anticipating the publication of the Government's policy paper on genetics, which was announced in early 2002.

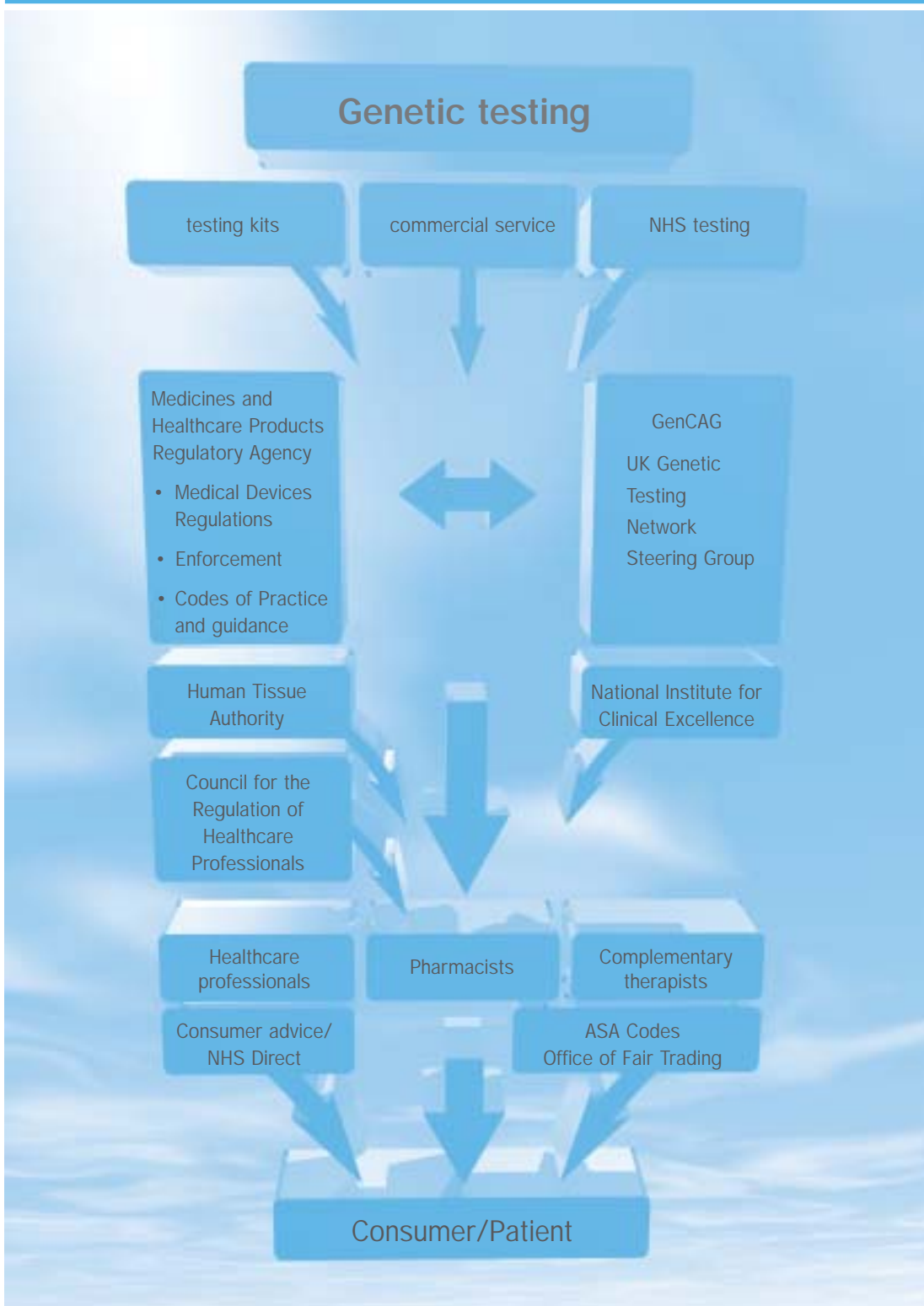
3.39 **The Government is already making some big changes to the legal and regulatory framework that will have an effect on direct genetic testing. The following proposals are intended as a framework that can guide those bodies that are responsible for regulation in this area:**

3.40 We conclude that the creation of the **Medicines and Healthcare Products Regulatory Agency** (MHRA) provides an excellent opportunity to develop an appropriate regulatory framework for direct genetic testing services, not least by promoting better voluntary standards across the industry. We do not wish to suggest that the regulatory regimes for medicines and medical devices should be aligned or merged.

3.41 In Part 2 we review the current requirements under the Medical Devices Regulations that affect genetic testing kits and commercial genetic testing companies. From December 2003 companies will need to CE mark in vitro diagnostic devices to show that they comply with the essential technical requirements in the relevant EU Directives. We believe that the regulatory system for marketing some genetic test kits and, in turn, some aspects of commercial genetic testing services under the new Regulations should be clarified among the industry groups, as at present there appears to be some confusion. We concluded that any genetic testing for predictive health



Figure 2: How the regulatory framework might look



purposes, pharmacogenetics, lifestyle testing or carrier testing should be considered to be a measurement of a 'physiological function' and part of the 'diagnosis, prevention, monitoring, treatment or alleviation of disease'. It follows from this that such tests fall under the regulations and that kits and commercial testing laboratories must meet certain minimum standards. We would welcome assurances that the MDA (and MHRA) will continue to work on clarifying the interpretation of the EU directives in relation to commercial genetic testing and, where appropriate, promote the voluntary adoption by industry of more stringent arrangements for oversight and advice.

Review of genetic tests

3.42 There appeared to us to be no specific regulatory oversight of the scientific or clinical validity of genetic testing or other commercial diagnostic services. Using our analogy, this appears to be akin to a medicines regulatory system that only requires medicines to be pure and safe without assessing whether they are effective at curing a disease. This is the approach currently taken for homeopathic remedies (but one which is gradually changing). In our wider work, we have considered the implications of pharmacogenetics for medicinal licensing. In considering applications for medicines which are prescribed on the basis of a pharmacogenetic test, regulators such as the MHRA may be required to consider some of the wider aspects of the genetic test associated with the medicines, such as accuracy, utility and possible wider ethical issues.

3.43 We conclude that there is evidence to suggest that all sides, including the industry, would welcome clarity on the arrangements for considering the scientific and clinical value of predictive genetic testing. We feel that the MHRA should consider how to extend the regulatory system for in vitro diagnostic devices in order to address wider aspects such as scientific quality and clinical utility. We formed the impression that the MDA and MCA feel that they are not empowered to consider the wider aspects such as scientific or clinical validity of tests or their ethical implications. We do not believe that they are statutorily barred from such considerations, but if so, we would hope that the Government would address such restrictions in setting up the MHRA.

3.44 We have considered various factors that need to be considered when judging the merits of a particular genetic test. These include:

- the technical performance of the test (its accuracy and reliability);
- the evidence supporting the interpretation of the test and the claims made for it (its scientific and clinical utility) which might include the value of the treatment or prevention options (such as medicines or dietary supplements), and
- the wider service quality of the organisation conducting the test, the arrangements for confidentiality, retention of data, the level of customer support and the marketing and advertising of such tests.

3.45 In time, we believe that all genetic tests should be subject to some form of oversight. We have noted that the **UK Genetic Testing Network** of GenCAG will be considering the question of the scientific and clinical validity of genetic tests for NHS use. This mechanism might be extended, or adopted in parallel, for genetic tests that are to be supplied privately, either by private medical referral or direct to the public. We broadly agree with the view that there needs to be some independent mechanism to



consider the scientific and clinical validity and utility of any genetic testing service. We feel that any commercial genetic testing service that is not ‘approved’ by established NHS mechanisms should be subject to an appropriate review, overseen by the MHRA, to consider the analytical quality of the tests, the route by which such tests might be offered.

3.46 In our view there needs to be a coherent review framework that can cover genetic tests across a spectrum that may range from the highly penetrant mutations in genes of large effect to those with loose associations with disease and those that are of relatively little health consequence. The outputs from studies like the UK Biobank may result in a large number of specific polymorphism which in combination may have some clinical utility. There will, potentially, be a significant delay while the evidence necessary to validate such tests is collected, and the regulatory agency is reviewing the dossier and reaching an opinion.

3.47 We feel that the work of the UK GTN will provide a valuable basis for developing appropriate review criteria. Tests with a high ‘informational impact’ and with significant health outcomes seem to demand a higher standard of proof than trivial ones. The challenge, as we found when considering the Sciona service, will be to set the criteria at an appropriate level for each test, particularly where there is little established scientific or clinical evidence to support claims made for the clinical value of a particular test. This delicate balance in the ‘risk-benefit’ of genetic tests is something that we would wish to monitor, working in conjunction with the UK GTN Steering Group and other interested bodies. It is also something that we believe will benefit from appropriate international liaison.

A new Human Tissue Authority

3.48 The review of the legislation on human organs and tissue – as indicated in the *Human Bodies, Human Choices* consultation – may provide for statutory regulation in the areas of consent, storage and disposal of most types of human tissue. There may also be a new **Human Tissue Authority** that will be responsible for the licensing of the activities of some genetic testing laboratories. We believe that there are some potential opportunities presented by the proposed Human Organs and Tissue Bill and we trust that our report will be relevant to the future passage of the Bill.

Industry self-regulation

3.49 We noted concerns by some of those we consulted that there is no specific external regulation of wider aspects of genetic testing services (such as arrangements for consent and counselling, confidentiality, storage of samples and communication of results). We recognise that commercial companies often meet or exceed the relevant quality assurance standards for sound commercial reasons.



3.50 We welcome the existing voluntary arrangements within the MCA and MDA to encourage manufacturers to adopt more stringent controls in some areas. We offer some general observations to Government and through them to the MHRA, the industry and healthcare professional bodies. We suggest that all parties consider a variety of statutory and non-statutory methods, such as Codes of Practice or Professional Ethics, aimed at achieving a high level of consumer protection.

3.51 We would like to emphasise that our discussions with the **UK Office of Fair Trading** (OFT) suggested that regulatory bodies like the MHRA could oversee stringent Codes of Practice. The OFT have a mechanism to ensure that all such Codes are subject to proper compliance checking and monitoring. We therefore do not automatically consider that our proposals will require statutory legislation, as this will be something for the regulators and others to consider further.

Professional self-regulation

3.52 We believe that any controls on industry at the time that a commercial service is made available should be supported also by controls on health professionals who may be involved in providing access to such tests or in providing advice on the interpretation of a test. We considered two different groups in our review – pharmacists and nutritional therapists.

3.53 We encourage the Royal Pharmaceutical Society of Great Britain to continue to develop robust professional standards for genetic testing. An individual pharmacist should have to meet the relevant professional guidelines covering their knowledge and skills in genetics and have appropriate facilities such as private consulting rooms. The role of pharmacists in providing access to some genetic testing services seems to us to be in line with current developments aimed at improving partnerships between pharmacists and GPs.

3.54 Other professional groups, for example genetic counsellors or nurse practitioners, have developed suitable professional codes and therefore should potentially be eligible to offer certain genetic testing services.

3.55 We have some reservations at this stage about allowing other groups of health professional or therapists to provide predictive genetic tests. On the basis of the evidence we gathered, we feel that there needs to be additional work to improve training and accreditation in genetics before groups such as nutritional therapists should actively offer genetic testing services for diet and lifestyle. We conclude that, at this stage, diet and lifestyle genetic testing services should be restricted to qualified public health nutritionists or to State-registered dieticians who have adequate knowledge and accreditation in human genetics.

3.56 We believe that there may be a role for the new over-arching bodies established to oversee aspects of professional self-regulation. The Health Professions Council will be increasingly involved in the professional regulation of the ‘allied health professions’. When it begins work in April 2003, the new **Council for the Regulation of Health Care Professionals** will have oversight of the self-regulatory role of the main professional bodies such as the General Medical Council, the Health Professions Council and the Royal Pharmaceutical Society of Great Britain. We suggest that the new



Council and other regulatory bodies should take note of our report and considers its role in promoting high standards of professional self-regulation.

Advertising and marketing of genetic testing services

3.57 However well we regulate matters in the UK, consumers will still have to make choices and will still be exposed to advertising on the Internet. We therefore conclude that an essential corollary is consumer education and the provision of accurate and impartial information in a variety of ways.

3.58 The recent public awareness of the advances in modern genetics could be misused as a powerful marketing tool by unscrupulous companies in support of misleading claims or in order to promote the buying of expensive and unnecessary dietary supplements or remedies. We have ample evidence that the public feel that some vulnerable individuals (the young, elderly or the ‘worried well’) need to be protected.

3.59 In our view the Advertising Standards Authority Codes of Practice and the OFT campaign on misleading health advertisements should be carefully studied and provide a very useful basis for controlling advertising and for a public education campaign on direct genetic testing services. The industry should rise to the challenge and only make responsible claims and on the accepted principles ‘legal, decent, honest and truthful’. Consumers also need to embrace a healthy degree of scepticism, perhaps summed up by the OFT slogan *“if it sounds too good to be true, it usually is!”*

3.60 We have noted developments in the USA regarding the marketing and promotion of commercial genetic testing services that are to be accessed via medical practitioners. We tend to agree that such advertising should be discouraged. One approach to this would be for the Committee on Advertising Practice to amend the Codes to require adverts for direct genetic testing kits and services to be directed only at the relevant health professionals. The exception would be where a direct genetic test service was ‘approved’ for wider provision.

Consumer awareness

3.61 We believe that funding should be made available to an independent consumer body in order to prepare specific and impartial information on any direct genetic testing services that are marketed. The aim should be to equip customers with sufficient information to judge the relevance of that service for them. Ideally, there should eventually be awareness of the scheme in a similar way to the established BSi ‘kitemark’ or CE mark or the industry-sponsored ‘ABTA’ and ‘ATOL’ schemes for holidays and travel.

3.62 In addition, we would like to see a broader Government effort to educate consumers about genetic testing and, in particular, predictive genetic testing. Such a campaign will be of wider value to ensure that any marketing or publicity around commercial genetic testing services, that is those that are available via referral by GPs or other health professionals, does not impose undue burdens on NHS resources. We look forward to seeing the proposed national strategy on patient information that is being developed by the Department of Health. We also suggest that advice and guidance material on direct genetic testing services is made available on established UK Internet sites, such as NHS Direct and the HGC website.



International liaison

3.63 Given the potential for Internet-based genetic testing services, we also recommend continuing liaison with the relevant international agencies dealing with genetic testing. In particular, we note the activities of the US Food and Drug Administration that is considering regulation of direct tests in the light of the SACGT report. The Australian Law Reform Commission is also soon to make proposals for including 'DIY' genetic testing within the scope of existing regulatory structures. We also highlight the importance of working with EU bodies, the Council of Europe, and professional and industry groups to encourage common approaches to direct genetic tests.

3.64 We accept that our report contains some potentially far-reaching proposals in an area where both the industry and the regulatory bodies are still developing. We will keep the situation under review and, in addition, propose a workshop or conference to consider progress in the light of the Government response to this report.

Other DNA testing services

3.65 There are some types of genetic testing (as we define it) which will almost certainly take place outside of any medical consultation. In particular, the use of 'DNA fingerprinting' techniques to establish family relationships (such as paternity testing) or ancestry (genealogy testing) are potentially large markets for commercial DNA testing. We have not considered in detail the implications of our report for these types of testing, but we intend conducting a short additional review of the current arrangements for paternity testing and genealogy testing.

3.66 We are also concerned about the potential development and marketing of genetic tests for behavioural traits, such as IQ or specified learning disorder like dyslexia. We have commented on the thorough report by the Nuffield Council on Bioethics and we believe that there are many serious implications of such tests. Any direct genetic tests for behavioural traits will require careful prior consideration of the clinical and scientific validity of the test and any related interventions. Such services should only be offered along with extensive and independent counselling and support. We feel that the ASA and OFT should pay careful attention to the advertising and marketing of any such tests and the claims that are made. We will also continue to monitor developments in this area in the UK and elsewhere.

**Human Genetics Commission
March 2003**



Glossary of some technical terms and abbreviations

ABPI	Association of the British Pharmaceutical Industry
ACGT	Advisory Committee on Genetic Testing
ASA	Advertising Standards Authority (an industry body that regulates voluntary Codes on advertising)
ASR	Analyte Specific Rule (a regulation by the FDA)
BIA	BioIndustry Association
BIVDA	British In Vitro Diagnostic Association
Carrier	A person who has one copy of a mutated gene causing an inherited recessive disorder. They are not affected but may pass the mutated gene onto their children
CE mark	A mark displayed on an article that complies with relevant EU product regulations or standards
CHAI	Commission for Health Audit and Inspection
CLIA	Clinical Laboratories Improvement Act 1988 (USA Federal legislation)
Cystic fibrosis	An inherited disorder which is commonly associated with a build up of mucous in the lungs and other systemic effects
Democs	Deliberative Meetings Organised by Citizens (see Annex 2)
DNA	Deoxyribonucleic acid – the chemical carrying the inherited aspects of a human cell
EU	European Union
FDA	The USA Food and Drugs Administration
GenCAG	Genetics Commissioning Advisory Group
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)
GIG	Genetic Interest Group



GMC	General Medical Council, the professional regulatory body for doctors
GP	General Practitioner
GSDL	Great Smokies Diagnostic Laboratories
Haplotype	A set of linked genetic markers that are generally inherited together
HGC	Human Genetics Commission
IVD	In vitro diagnostic device – defined by the In Vitro Diagnostic Devices Directive (98/79/EC) – the IVD directive
MCA	Medicines Control Agency which regulates licensing, manufacture and sale of medicines and some aspects of herbal and homeopathic remedies
MDA	Medical Devices Agency which regulates medical equipment including artificial implants and diagnostic machinery
MHRA	Medicines and Healthcare Products Regulatory Agency, formed in April 2003 from the merger of MCA and MDA
NEF	New Economics Foundation
NHS	National Health Service
NICE	National Institute for Clinical Excellence
NIH	The USA National Institutes of Health
Nutrigenomics	A developing scientific discipline of using the knowledge of individual metabolism and genotype to determine diet
UK GTN	United Kingdom Genetic Testing Network, a sub-group of GenCAG which coordinates the NHS network of genetic testing laboratories
OFT	Office of Fair Trading
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
PSA	Prostate specific antigen, a diagnostic indicator of prostate cancer
RNA	Ribonucleic acid – a chemical involved in translating genetic information into structural or metabolic proteins
SACGT	The US Secretary's Advisory Committee on Genetic Testing
SNP	Single nucleotide polymorphism; 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



Working Group on Genetic Testing Services supplied Direct to the Public

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
Stephen Bain (from Jan 03)
Ruth Evans (to Nov 02)
Hilary Harris
Sandy McCall Smith

Patrick Morrison
Hilary Newiss
John Sulston
Veronica van Heyningen
Geoff Watts

The Working Group met in June 2002 to discuss and agree the written consultation document. It met a further six times between September 2002 and January 2003 to consider the responses to the consultation and prepare the final report for the Commission.

The Working Group was disbanded in March 2003 on completion of this report.



The consultation process

We decided on a varied programme of consultation to inform our review of direct genetic testing services. Details of how to obtain copies of the various responses and reports mentioned below are given in Annex 3.

- **Consultation document** – in July 2002 we published a consultation document, including a summary version and we received 65 written responses.
- **Evidence-gathering meetings** – while the consultation was underway, we held a series of evidence-gathering meetings with key groups and individuals (see Box 1). This enabled the HGC Working Group to gain an understanding of the wider aspects of the regulatory climate or to hear first-hand the view of industry, professional and patient groups and consumers.
- **Contributions from British Embassies** – a number of British embassies outlined the situation with respect to direct testing in the country in which they were based.
- **Consultative Panel** – the HGC Consultative Panel discussed this on 16 July 2002, when one of the afternoon roundtable discussions focused on the issue.
- **Plenary meeting** – our meeting in Belfast on 11 September 2002 included time for a discussion with the audience on direct testing.
- **Wider public consultation** – recognising that the market for direct genetic testing services will be largely driven by consumer demand, we felt it was important to seek views from consumer organisations but more importantly from the wider public. As well as our written consultation document, we conducted a series of focus groups, an internet-based survey via YouGov, and a series of public discussion meetings. More detail about each of these activities is given below.

Focus groups:

We commissioned People Science and Policy Ltd to run a series of focus groups. Eight groups were conducted in November 2002. The groups were set up to cover a range of ages to make sure we heard from younger and older people and those with younger and older children, and were held in Reading, Watford, Leeds and Birmingham. In summary, the results told us:

- **Use of tests** – there was no awareness of genetic tests sold direct to the public among the groups.



People thought that the main reasons for others to use such tests would be speed, convenience and confidentiality, however they saw very little need for people to use such tests in the context of a free NHS.

Most also did not feel that the test results would be of particular use to them.

The medical profession is seen as the best source of information and advice on genetic tests and people would overwhelmingly turn to their GP for advice if they had taken a test themselves.

- **Credibility** – doctors were also seen as the most credible source for genetic tests followed by pharmacists, and mail order and the Internet are very much less credible.
- **Counselling** – some felt that individuals are not equipped to deal with the results of genetic tests and would need medical input to understand them. Others felt they should have access to such information if they wanted it.
- **Advertising** – all groups were concerned about those they saw as vulnerable (the elderly, those who live alone and the worried well) and felt they should be protected against companies directing their advertising at them.
- **Policy options** – People aren't aware enough of the existing regulations to give an opinion on these. Views were divided between support for strict regulation and support for the free market, but self regulation by companies was not a well supported option.

Survey of public attitudes

We commissioned YouGov to carry out an internet survey of public attitudes to direct genetic testing. This was carried out between 27 December 2002 and 5 January 2003 with a nationally representative sample of 2510 people. The data has been analysed using a number of sub-groupings, including age, gender, region, profession, income, history of hereditary problem, level of genetic knowledge and educational results. In summary, the results told us:

- A third of respondents have bought a health testing kit to carry out at home, but very few have bought a personal health test via mail order or internet (1% of the sample for each).
- While a third of people said they would use a genetic testing service, only a fifth of people said they were likely to want to use a genetic home testing kit.
- Unsurprisingly people are more likely to use a genetic home testing service if it is offered by their doctor (an average of 81% of people were more likely to use a range of genetic home testing services if their doctor recommended them, compared to 49% if friends/family had used the service and 15% if they had seen/heard an advertisement).
- Most people would prefer to receive information about genetic tests and the results of tests from their doctor (63% and 67% respectively). The second most preferred option was to receive information or the results of a test in a leaflet or letter (14% and 12%) and the third was to receive information or results 'At a face

to face appointment with a doctor who works for the testing company' (7% and 8%).

- People generally consider GPs or NHS specialists to be the most appropriate way to access genetic tests. The degree to which the public consider them to be the most appropriate source depends on the nature of the genetic test, increasing for tests where the results can have serious consequences for an individual's health or emotional state (88% for tests for serious disorders and 85% for carrier testing).
- People were asked about lifestyle tests, paternity tests, genealogy tests and tests for serious disorders and for each test 'a GP or NHS specialist' was the top answer from a list of five.
- Generally people trust prescription services and Government approved services. From a list of 5 options (excluding Don't Know) that might make people trust a genetic testing service, respondents identified 'A test that is available on prescription from a doctor' as the one that would inspire most trust in the service and results.
- The public doesn't trust companies to self-regulate; 'A test that is provided by a company that abides by a detailed voluntary code of practice' was the option fewest people said would make them trust a genetic testing service and the results it gives.

Democs

We also worked with the New Economics Foundation (NEF), who developed Democs (Deliberative Meetings Organised by Citizens) as part of its Democracy and Decision-Making programme. Democs were developed as a new way for people to come together, absorb information, come to a view and seek common ground. NEF ran a series of public meetings to tie in with HGC's review, and HGC Consultative Panel Members were also invited to attend.

The project's aim is to allow people to run their own focus group-style events. A kit is prepared including cards about direct genetic testing aimed at sparking ideas and providing basic factual information; participants use the cards to aid discussion and vote on their own position before and after their deliberations.

NEF organised six events between the 7th and 17th of January 2003, three in London and one each in Bristol, Birmingham and Edinburgh. A total of 47 people attended the events, including 14 Panel Members. NEF prepared a report of the meetings that gives a description of the conclusions the groups came to and the arguments that swayed them. The people who came were clearly self-selected and unrepresentative and the report is not intended as a guide to what the population as a whole would think. In summary:

- People were given possible regulatory options and asked which they supported – the option of no strict regulation was almost wholly rejected, three quarters of the participants were prepared to support strict regulation and the most heavily supported position was one of voluntary regulation with restrictions on types of test.



- The arguments that led to these policy positions have been grouped, in order of ‘popularity’:
 - Seven arguments – a third of the total – concerned the impact of a test on the recipient and her/his family etc. This is what pushed many people towards supporting considerable regulation.
 - Four arguments concerned the rights of individuals.
 - Another group of arguments differed by being concerned not with the individual taking the test but with the overall effect on resources.
 - At least two groups pointed out that this involved the wider issue of the future of the NHS.
- A number of dilemmas were identified with which people would need help. As one group noted, ‘On an ethical level, who should make the decisions about the result for a third party? For example, if an unborn child is discovered to have a certain disease, who should decide whether or not to terminate the pregnancy, the mother or the father? The State (could it reach that point?)’
- Two clusters of arguments concerned the quality and accuracy of tests.
- Only one group produced an argument explicitly about regulation, although issues such as quality are closely connected.

People were also asked to fill in a feedback form and the report includes the comments of all those who replied. The questions asked about dilemmas people identified, the arguments which particularly influenced their views, anything people would like HGC to take into consideration and what was most satisfying or valuable and what was less than satisfying or disappointing about the Democ. In general, people seem to have found these interesting and rewarding events.

Evidence

We drew on a wide range of evidence during our review and in common with previous reports we have made this available in full on the HGC website www.hgc.gov.uk/genesdirect/

Paper copies of the evidence are available via the HGC Secretariat. If you would like further information please telephone 020 7972 1518 or email hgc@doh.gov.uk.

Consulation document

Human Genetics Commission. The supply of genetic tests direct to the public: A consultation document (July 2002). This can be found on the website.

The list of those who submitted written responses to the consultation document follows. Copies of the responses can be found on the website (except where we have been asked to treat the responses as confidential).

Organisations

Advertising Standards Agency/Committee of Advertising Practice	Christian Centre, Nottingham
Alliance Unichem (Moss Pharmacy)	Christian Medical Fellowship
Association of British Pharmaceutical Industry	Church of England Board of Social Responsibility
Astra Zeneca	Consumers' Association
Bevan Ashford	Contact A Family
BioIndustry Association	Diabetes UK
Boots the Chemists	DTI Policy advisor on Internet regulation
Breakthrough Breast Cancer	European Bioethical Research
British Association for Nutritional Therapy	European Commission Health and Consumer Protection Directorate-General
British Humanist Association	Genetic Interest Group
British In Vitro Diagnostics Association	GeneWatch UK
British Medical Association	GlaxoSmithKline
British Society for Human Genetics	Health Technology Board for Scotland



Human Genetics Alert	Royal College of Physicians of Edinburgh
Information Commissioner	Royal Pharmaceutical Society of Great Britain
McFarlane Valentine BioConsulting (MVBio)	Sciona
Medical Research Council	UK Forum on Genetics and Insurance
National Council of Women of Great Britain	US Food and Drug Administration
Nuffield Council on Bioethics	US National Society of Genetic Counsellors
Occupational Health Advisory Committee/Health and Safety Executive	US Secretary's Advisory Committee on Genetic Testing
Royal College of General Practitioners	Wellcome Trust

Individuals

Ms Janey Antoniou	Ms Alice Maynard
Dr Rosemary Boothman	Ms Audrey McLaughlan
Dr Mark Brennan	Mr Barry Pearson
Mr Roger Bull	Ms Sarah Pettifor
Mr Colin Butts	Dr Sultana Saeed
Dr Paul Debenham	Ms Gill Wallace-Hadrill
Dr Ros Eeles	Mr Robert Walton
Dr Ian Ellis	Prof Martin Whittle
Mr Jonathan Gray/Ms Lindsey Prior	Welsh Senior Medical Officer
Ms Shirley Hodgson	Dr Ron Zimmern
Prof Theresa Marteau	

There were also two anonymous responses.



Evidence-gathering meetings

We have summarised the face-to-face discussions we had with a number of organisations and these are available on the website.

Advertising Standards Agency	Human Genetics Alert
BioIndustry Association	Medical Devices Agency
British Association of Nutritional Therapy	National Pharmaceutical Association
British In Vitro Diagnostic Association	Royal Pharmaceutical Society of Great Britain
Consumers' Association	Sciona Ltd
Dr Paul Debenham (LGC)	Steering Group for National Network of Genetic Testing
Genetic Interest Group	UK Office of Fair Trading
GeneWatch UK	
Great Smokies Diagnostic Laboratories	

Contributions from British Embassies

British Embassies in Canada, Germany, Holland, Japan and US outlined the situation in their respective countries, and their reports can be found on the website.

Consultative Panel meeting and HGC plenary meeting

A note of the discussions at the Consultative Panel meeting on 16 July 2002, and after an HGC plenary meeting in Belfast on 11 September can be found on the website.

Wider public consultations

Reports of the focus groups, the YouGov internet survey, and of the Democs events can be found on the website.



Controls in other countries

4.1 We felt that it would be useful to consider the regulatory approach taken by other countries, especially those where clinical laboratory services are supplied by private companies. We sought views via the FCO Science and Technology Network, European Commission and by direct contact with relevant advisory bodies.

United States of America

4.2 We have paid careful attention to the 2000 SACGT report *Enhancing the oversight of genetic tests* and elsewhere we echo many of their findings and definitions about the requirements for oversight. The report also serves as an extremely useful summary of the US regulatory mechanisms. The report recommended, among other things, that the US FDA be involved in the regulation of genetic tests, whether they are packaged and sold as kits or provided as a laboratory service. We also note that SACGT has recently been replaced with a broader committee – the Secretary’s Advisory Committee on Genetics, Health, and Society. We look forward to future discussions with them about progress on the oversight of genetic tests.

4.3 The previous administration appeared to have accepted a role for the FDA in oversight of tests. However, the detailed implementation of the recommendations, including the development of criteria for oversight, is still continuing. Our understanding of the current regulatory position from the FDA is that the Food, Drug and Cosmetic Act (FFDCA) require some device manufacturers to notify FDA of their intent to market a medical device. The 1976 Medical Device Amendments to the FFDCA established three regulatory classes for medical devices. Genetic tests packaged and sold as kits are subject to regulation by the Food and Drug Administration. These kits require pre-market approval by FDA for safety and effectiveness.

4.4 Many clinical laboratories advertise tests direct to the public as a service, particularly ‘home brew’ tests developed at the lab with commercially available reagents. While FDA acknowledges it may have authority under the Medical Device Amendments to regulate genetic tests marketed as services, it has not yet done so. They have recently made public plans to move forward with a ‘Notice of Proposed Rule Making’ for incremental regulation of all ‘home brew’ tests according to their risk. This goes somewhat broader than the recommendations made by the SACGT and eliminates the potential for ‘genetic exceptionalism’ by singling genetic testing out for greater oversight.

4.5 The FDA also noted that ‘home brew’ tests offered directly to consumers are actually illegal under the Analyte Specific Reagent (ASR) rule, although there are some potential loopholes. The FDA is also concerned about the difficulty of regulating Internet testing services because of the difficulty of controlling activities outside of the USA jurisdiction.



4.6 More broadly, the 1988 Clinical Laboratory Improvement Act (CLIA) established quality standards for all US diagnostic laboratory testing to ensure accuracy, reliability and timeliness of patient test results. The stringency of CLIA requirements depends on the complexity level and speciality to which tests are assigned. Although the current CLIA regulations cover all genetic testing, we understand that at present the only specific requirements for genetic tests under CLIA are for cytogenetic tests (e.g. for chromosomal abnormalities like Down's syndrome).

4.7 Under CLIA, clinical laboratories must demonstrate analytical validity of their tests but there is no statutory or regulatory requirement for them to establish the clinical validity and clinical utility (the value for prevention or treatment) of clinical laboratory tests. There is also no requirement for post-market surveillance of these tests.

Canada

4.8 At the Federal level genetic tests marketed as a kit must be approved by Health Canada. There appears to be no specific regulations covering testing services, or covering services provided by companies in other countries (particularly the USA). At the provincial level, some, for example Ontario, have expressed concerns over direct genetic testing services and have recommended that Federal standards for approval and review of such at-home tests should be carefully examined and monitored to ensure that they adequately protect the public¹¹.

4.9 Although direct to consumer advertising is prohibited in Canada for prescription medications and professional services, such advertising does penetrate the Canadian market via USA broadcast media and the Internet. Direct to consumer advertising may well influence the adoption of and demand for genetic services. Ontario has raised some concerns about the appropriateness of genetic testing with no formal requirement of patient education or risk management advice. They have also suggested the establishment of a Canadian equivalent of the Human Genetics Commission to aid in the provision of a co-ordinated approach across jurisdictions.

Australia

4.10 The Australian Law Reform Commission (ALRC) has been conducting a major review of the law relating to human genetic samples and information and we have held some informal discussions with Professor David Weisbrot who is leading the review and with Mr Malcolm Crompton, the Federal Privacy Commissioner. We have been impressed with the thoroughness with which the ALRC have conducted both the initial consultation and the subsequent discussion phase.

4.11 The Discussion Paper 66, published in August 2002 covers several relevant aspects. In particular, we noted the concerns about the possibility of non-consensual testing, and the recommendation for a similar offence to the one that we have recommended. We also noted that they are considering recommending the establishment of the Human Genetics Commission of Australia (HGCA).

¹¹ Genetics, Testing & Gene Patenting : Charting New Territories in Healthcare. (2002) Ontario Ministry of Health and Long-Term Care (see www.gov.on.ca/health)

4.12 In relation to direct genetic testing services, the Discussion Paper says:

“To date, medical practitioners have been the primary ‘gatekeepers’ of genetic testing and the information derived from it, at least for clinical purposes. Increasingly, however, there are other paths by which genetic testing may be accessed. These include direct access to testing services provided by laboratories and over-the-counter or home use genetic testing kits, including for parentage testing. The Inquiry proposes that these forms of ‘do-it-yourself’ genetic testing should be regulated by the Therapeutic Goods Administration, with advice from the HGCA.”

We look forward to seeing the outcome of their detailed report that is due to be published in March 2003.

Japan

4.13 In Japan, at present there is no Government enforced regulatory regime to control the supply of genetic tests direct to the public. There are voluntary guidelines for genetic testing for clinical genetic diagnosis that are issued by the relevant learned societies. Among other things, these seek to prohibit advertisements to the general public. In 2000, the Japan BioIndustry Association submitted a detailed report to Government, asking it to introduce legislation banning all genetic tests unless they were requested, administered and feedback provided by a medical doctor. The Government has so far not responded.

4.14 In Japan a few private companies offer a broad range of genetic tests, most of which operate via the Internet despite the guidelines. They offer a variety of genetic tests to predict for example susceptibility to osteoporosis, some cancers and Alzheimer’s Disease. Although these companies were registered in Japan, the likelihood is that the tests themselves are carried out in the USA. We heard of a new small company, DNA Bank Inc, is providing health and lifestyle advice based on DNA test results. It envisages health instructors, based in up to 3000 sports shops or health clubs, who will analyse, based on the results of DNA tests, a client’s susceptibility to become overweight for example and will advise on how to improve lifestyle (exercise and diet).

4.15 There is increasing concern amongst the Japanese genetics community because the guidelines are not enforceable and therefore are increasingly being ignored. They also fear that because the accuracy and efficacy of tests have not been officially approved or assessed there is a potential risk that clients are could be misled.

European Community controls

4.16 We were particularly interested to consider the approach taken in European Union (EU) countries which are obliged to implement relevant EU legislation. There have been a series of Directives aimed at securing a Single European Market for medical and other products. Of particular relevance here are the Medical Diagnostic Devices Directive (93/42/EEC, as amended) and the In Vitro Diagnostic Devices Directive (98/79/EC) – the IVD directive.



Netherlands

4.17 We had an opportunity to discuss the early stages of our review when we met the Dutch Platform for Medical Technology in June 2002. We understand that in 1999 the Dutch Council for Health and Care decided that self-testing kits, such as those for determining carrier status, did not need special regulation. The Government took the view that the IVD directive would cover the quality requirements and that there was no reason to interfere with the user's 'right of self-determination'. In March 2002, Government regulations (Regulering In-Vitro Diagnostica) introduced a form of consumer protection. Self-testing products, potentially including genetic tests, remain available to the public. Some, however, may only be obtained through a GP, specialist or chemist.

Germany

4.18 There is still a debate in Germany about genetic testing in general rather than services that are available directly to the public. The legislative framework is not fully developed and there are plans in the new government coalition agreement for legislation on genetic testing and the use of genetic data. One area of concern noted in Germany is the potential for non-consensual genetic testing – for health or paternity purposes.

4.19 The quality and reliability of genetic tests are covered by the Medicinal Products Act, which implements the IVD Directive. There are particular concerns amongst the German medical community and some industry groups about the potential for unregulated testing via the Internet.