



Human  
Genetics  
Commission

# *More Genes Direct*

A report on developments in the availability,  
marketing and regulation of genetic tests  
supplied directly to the public



December 2007





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## Contents

Foreword		3
Summary		5
Chapter 1	Background	7
Chapter 2	Recent developments	11
Chapter 3	Responding to developments	15
Chapter 4	The role of the HGC	21
Chapter 5	Consolidated recommendations	23
Glossary		27
Appendix 1	Meeting participants	29
Appendix 2	Companies offering genetic testing services in the UK	31
Appendix 3	UK Government response to the HGC's <i>Genes Direct</i> report	39
Appendix 4	Correspondence regarding the advertising of genetic tests	41





## Foreword

It is over four years since the Human Genetics Commission (HGC) published its report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. Those years have seen a consistent increase in the number of genetic tests on the open market. Almost every time the HGC meets, we hear about a new test becoming available and, simultaneously, about concerns regarding the test's efficacy, utility or its implications for individuals and their families. It is not yet possible to say whether we are on the verge of an explosion in direct-to-public genetic testing or whether we should expect merely a steady increase. However, in the absence of any significant regulatory controls, the future of direct-to-public genetic testing is, for now, largely in the hands of commercial test providers: the pharmaceutical companies, their marketing departments and PR agents.

In particular, we are now seeing a burgeoning cottage industry in so-called 'lifestyle' tests together with the regimens, dietary supplements and self-administered medications that they are claimed to indicate. Some of these tests are relatively innocuous. There may even be benefits in that they provide reassurance or a sense of empowerment, or encourage the adoption of a healthy exercise regime. We might, then, simply counsel the buyer to beware and leave it at that, were it not for the fact that, in the same marketplace, other tests are available with potentially more serious implications. Tests that claim to predict the onset of disease or indicate a heightened risk of serious conditions, or, alternatively, to offer peace of mind and the promise of a long and active retirement, can significantly influence choices that profoundly and enduringly affect an individual's health. These, we think, need to be provided in the context of proper consultation, where their implications can be discussed and managed. And even 'lifestyle' tests, for which neither the 'diagnosis' nor the prescribed 'treatment' stand out clearly from the normal ranges of genetic variation and modern behaviour, where they are not provided with adequate or appropriate advice, can also have a harmful effect, for example, heightening anxieties about health or encouraging a complacent disregard for the effects of an unhealthy lifestyle.

The HGC is not a regulatory body or a licensing authority: it would not be appropriate for us to single out particular companies or tests. But as the Government's advisory body in developments in human genetics, the HGC is frequently petitioned by individuals, interest groups and professional bodies, and audience members at our regular public meetings, to bring about consistent oversight of the genetic testing marketplace. As our recommendations show, we are sympathetic to these concerns. In the coming weeks and months we intend to work with test providers and official bodies in the UK and at a wider European level to try to ensure effective oversight of genetic tests supplied directly to the public (the subtitle of our original *Genes Direct* report) and to develop fair but robust approval mechanisms, codes of practice and marketing guidelines.



We think the recommendations in this report are a proportionate and reasonable approach to the current situation as we perceive it developing, and we look forward to the response of, among others, the Government, official agencies, test providers and consumer groups to our proposals.

A handwritten signature in black ink that reads "John Sulston". The signature is fluid and cursive, with the first name being more prominent.

Sir John Sulston  
Acting Chair, Human Genetics Commission  
November 2007





## Summary

In March 2003, the Human Genetics Commission (HGC), the UK Government's advisory body on the legal, social, ethical and economic impact of developments in human genetics, published the report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. The report made a series of recommendations about how to develop the framework for the regulation of genetic tests in order to address what it saw as three key concerns:

- the danger that the public might receive misleading medical advice as a result of companies overstating the role of genetics in common complex diseases
- the difficulty of ensuring informed consent when tests are offered direct to the public
- the impact on NHS resources if patients were to seek advice from their doctors before or after tests, or if patients were to require confirmatory testing within the NHS.

The HGC's findings exposed the current patchwork of regulation governing commercial genetic testing in the UK, from consumer law to professional codes of conduct, and made recommendations about how these different mechanisms might be improved. The recommendations focused heavily on **predictive** genetic tests. The report recommended that predictive genetic tests should only be available via a consultation with a doctor and, like prescription medicines, should not be advertised directly to the public. The report stressed the need for an independent system of pre-market review to consider the scientific and clinical validity and clinical utility of genetic testing services and to determine whether they ought to be offered directly to the public. Where tests were to be offered directly to the public, the report concluded that a code of practice would assist in ensuring that consumers received a high standard of service. The report also recommended funding for educational initiatives to improve public understanding of genetics and the development of impartial sources of information on genetic tests.

In drawing together the 2003 report, the HGC concluded that although at the time there were few genetic tests being offered to the public, this situation could change rapidly within the next few years. The report therefore finished by saying that because the Commission had made "far-reaching proposals in an area where both the industry and the regulatory bodies are still developing", it would continue to monitor developments and hold a workshop to review the Government's response to its recommendations.

In January 2007, four years after the publication of *Genes Direct*, a meeting was held with the aim of reviewing the original recommendations, identifying regulatory gaps and making realistic and practical proposals for the HGC to take to the Government and also to European bodies where appropriate.

*Genes Direct* stated the HGC's commitment to ongoing collaboration with regulators within and outside the EU on the oversight of direct-to-public genetic tests. In addition to members of the HGC's own Genetic Services Monitoring Group, organisations with key roles in the international oversight of genetic testing, as well as key members of the Medicines and Healthcare products Regulatory Agency (MHRA) and other experts in the field, contributed to the meeting.



In preliminary discussions concerns were immediately identified relating to the levels of scientific evidence given by manufacturers in support of their genetic tests, the quality assurance processes of genetic test providers and the lack of independent consumer information available.

This report outlines the key aspects of this meeting and the agreed recommendations. These fall into three areas:

### Pre-market review

- The recommendation in *Genes Direct* that certain genetic tests are only offered by a suitably qualified health professional should be implemented.
- Medical genetic tests, which are covered by the In Vitro Diagnostic Devices (IVDD) Directive, are classified by the relevant authorities in the UK as ‘low risk’ and therefore exempt from independent pre-market review. This risk classification should be urgently reviewed.
- For those genetic tests that fall outside the IVDD Directive, such as so-called ‘lifestyle’ tests, an alternative regulatory mechanism should be established to provide reliable oversight.

### Quality assurance

- A code of practice relating to genetic testing services should be developed that will take into account the guidelines published by the Organisation for Economic Co-operation and Development (OECD) and other relevant international standards, e.g. EuroGentest should be developed.
- The development of the code of practice and its implementation should involve relevant stakeholders including government bodies, public bodies, charities and industry.
- The UK should engage with the Council of Europe and offer to participate in its work in this area.

### Advice and advertising

- Advertising for tests that are available only via medical consultation should be restricted to medical practitioners (i.e. no direct-to-public advertising).
- The Advertising Standards Authority (ASA) and the Office of Fair Trading (OFT) should consider enhancing the codes of practice for tests that may be marketed directly to the public.
- Existing web-based information sources should be used as a means of providing comprehensive and independent information for consumers. Test developers/providers should be encouraged to facilitate consumer access to this information.



## 1. Background

**1.1** In March 2003, the HGC, the UK Government's advisory body on the legal, social, ethical and economic impact of developments in human genetics, published the report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. The report was prepared in response to a specific request from UK health and science ministers in 2002 for a review of the provision of genetic tests offered directly to the public. Full details of the public consultation process that informed the HGC's deliberations, and the evidence and arguments supporting the recommendations made, are contained in the original report.

**1.2** In the report the HGC concluded that although at the time there were few genetic tests being offered directly to the public, this situation could change rapidly within the next few years. The HGC therefore recommended reviewing the take-up and continued relevance of the recommendations contained in *Genes Direct* after three years and examining the development, availability and regulation of genetic tests supplied directly to the public at that time.

### Regulation of direct genetic tests in the UK

#### Brief chronology

<b>1996</b>	Establishment of Advisory Committee on Genetic Testing (ACGT)
<b>1997</b>	Publication of ACGT's code of practice on genetic testing services supplied directly to the public
<b>1999</b>	ACGT's work subsumed within the newly established HGC
<b>2002</b>	HGC begins public consultation on the future of regulation
<b>2003</b>	Publication of <i>Genes Direct</i> , the HGC's report on the future of regulation
<b>2003</b>	IVDD Directive regulating diagnostic tests comes into force in the UK, enforced by the MHRA

**1.3** The regulation of commercial genetic testing in the UK began with the establishment of the ACGT in 1996. Its terms of reference required it to advise the government on developments in genetic testing, on the ethical, legal and social implications of genetic testing and "to establish requirements, especially in respect of efficacy and product information, to be met by manufacturers and suppliers of genetic tests".

**1.4** The ACGT produced a code of practice for genetic testing services supplied directly to the public which emphasised three principles: the need for informed consent, the provision of data on the validity and utility of tests to patients in a easily understood format and the importance of counselling. The code established a voluntary system of regulation, under which suppliers proposing to offer a genetic testing service directly to the public (or proposing an amendment to an existing service) were encouraged and expected to present their proposal to the ACGT prior to its introduction. The ACGT developed two further guidance documents – one for research ethics committees and



another on the provision of tests for late-onset disorders. Throughout its work, the ACGT consistently emphasised the importance of gathering and disseminating data on the accuracy and predictive value of tests (so, for example, in its report on late-onset disorders it recommended that the clinical validity of a test must be established before it enters clinical practice).

**1.5** In 1999 the role of the ACGT was taken over by the HGC in a reorganisation of the advisory and regulatory bodies for UK biotechnology. However, the primary role of the HGC was advisory rather than regulatory – to provide independent strategic advice to government. It was in this advisory capacity that the HGC was, in 2002, asked to review the regulation of direct-to-consumer tests and the *Genes Direct* report was the outcome of that review.

## Recommendations in *Genes Direct*

**1.6** In *Genes Direct*, the HGC defined a genetic test 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” and ‘direct’ genetic tests were defined as those tests “not offered as part of a medical consultation”. A further distinction was made between direct-to-public and direct-to-consumer tests:

- **direct-to-public (DTP)** tests are those which are provided via a non-medical intermediary, such as a pharmacist or alternative health practitioner
- **direct-to-consumer (DTC)** tests are those where the test is provided without an intermediary between the consumer and the test provider

**1.7** However, within the context of services currently provided in the UK it is often difficult to separate tests provided directly to the consumer from tests which form part of a medical or health promotion service provided directly to the consumer via an intermediary. Much of the work carried out by bodies such as the OECD and EuroGentest (which is discussed in more detail later in this report) is concerned with genetic tests that are used in medical services but marketed directly to the consumer. *Genes Direct* acknowledged this wider issue in identifying the need for some independent mechanism to consider the scientific and clinical validity and utility of any genetic testing service. If a company wants to provide a direct genetic test other than through a doctor, it concluded, it should be required to convince a regulator that the test is suitable. The establishment of the MHRA in the UK therefore seemed to provide an excellent opportunity to develop an appropriate regulatory framework for direct genetic tests before they are placed on the market. Furthermore, the UK Genetic Testing Network (UKGTN) has introduced arrangements for reviewing genetic tests, which also provide a useful basis for the oversight of direct genetic tests.

**1.8** The *Genes Direct* report suggested a framework to guide those bodies that are responsible for regulation to ensure that companies only market high-quality tests with good customer information and appropriate support, and that they do not seek to misrepresent the value of genetic information in their marketing, while stopping short of recommending that there should be a statutory prohibition of some (or all) direct genetic tests.



**1.9** To complement this it also recommended that there should be a well-funded NHS genetics service, supported by a genetically literate primary care service, which can properly manage and facilitate access to new predictive genetic tests that are being developed and address concerns about predictive genetic tests that may be carried out without appropriate medical supervision. This last concern is connected to the issue of consent and steps that should be taken to ensure that tests are carried out with the proper consent of the test subject.

**1.10** Since its publication, *Genes Direct* has had a substantial impact internationally in the ongoing debate as to how to regulate and control genetic tests. We know of colleagues and collaborators in Europe and Japan who are using *Genes Direct* in their discussions about the regulation of direct genetic tests. The remainder of this report considers what steps may now be necessary to update and follow through these recommendations.





## 2. Recent developments

**2.1** Since the publication of *Genes Direct* in March 2003, the HGC has been aware of a marked increase in the number of companies offering genetic tests and genetic testing services over the internet (see Appendix 2). In 1998, the IVDD Directive came into force, although it was not implemented by the UK until May 2003. Compliance with this legislation was initially overseen by the Medical Devices Agency (MDA). However in April 2003, shortly after the publication of *Genes Direct*, the MDA was amalgamated with the Medicines Control Agency (MCA) to create the new MHRA. *Genes Direct* anticipated that this could offer an opportunity to develop a more robust regulatory framework for direct genetic tests before they are placed on the market (pre-market review). However, the MHRA's powers under the IVDD Directive do not currently extend to pre-market review of the clinical validity and clinical utility of genetic tests, contrary to what the HGC anticipated in its earlier report. (The MHRA's reviews currently concentrate on the safety, quality and accuracy – the analytical validity – of tests.)

### Elements in the pre-market review of genetic tests

**Analytical validity:** accuracy of the test in identifying the biomarker

**Clinical validity:** relationship between the biomarker and clinical status

**Clinical utility:** likelihood that the test will lead to an improved outcome for the test subject

**Ethical, legal and social implications:** whether the use of the test involves additional considerations for individuals, certain groups or for society more generally

**2.2** The IVDD Directive operates on the principle of risk-based regulation where the level of regulation applied is intended to be relative to the risk of harm posed by a test. The principal reason that most genetic tests are not subject to independent pre-market review in the European Union is that they are classified as 'low risk', and therefore the manufacturer is not required to submit their technical file to the national Notified Body (the MHRA in the case of the UK). While the sampling device itself may pose a low risk in terms of its use, the complexity of interpretation and the nature of the information provided may merit a different risk classification. As noted in *Genes Direct*, the IVDD Directive has several categories of commercial test. These include:

- **general kits:** these require only self-certification
- **self-testing kits:** these are assessed to determine whether they should be classified in the high-risk category
- **'list B' kits or services:** this category includes some tests that might be classified as genetic
- **'list A' kits or services:** this category includes blood group testing and tests for some infectious diseases.





**2.3** Tests classified as ‘general kits’ are currently exempt from pre-market review and only genetic tests for three conditions or genotypes (phenylketonuria, HLA tissue type and Down’s syndrome) are classified as high risk, with most genetic tests currently available needing only self-certification.

**2.4** Tests offered by individual health institutions, described as ‘home-brew’ tests, also fall outside of the IVDD Directive. These are tests made by a single health institution and used on the same premises as their manufacture where the reagents and methods used are also legally owned by the institution. The IVDD Directive has a specific exclusion for tests of this kind.

**2.5** At the time of publication, *Genes Direct* welcomed the recently introduced proposals for reviewing genetic tests from the UKGTN. Since then, the UKGTN has taken on the role of monitoring genetic tests for single-gene disorders offered by the NHS. The UKGTN gene dossier system evaluates tests for these disorders and provides information on the clinical validity, analytical validity, clinical utility and ethical and legal implications. While it does not have a formal regulatory role, the UKGTN requires laboratories offering genetic testing through the NHS to submit a technical file to the UKGTN gene dossier, which helps to maintain a high level of quality assurance. Furthermore, laboratories within the UKGTN are required to be accredited through the Clinical Pathology Accreditation (UK) Ltd (CPA) system. The UKGTN agrees with the HGC’s view that genetic tests should form part of an integrated care pathway of health provision and therefore that necessary information must be provided to the patient and the results interpreted correctly, with relevant support being offered.

**2.6** In May 2003 the genetics White Paper *Our Inheritance, Our Future: Realising the potential of genetics in the NHS* was published. The White Paper set out an investment of £50 million to be invested in clinical genetics in England and helped to meet the *Genes Direct* recommendation for a well-funded NHS genetics service supported by a genetically literate primary care workforce. This funding has now all been invested and the HGC is awaiting the outcome of the Department of Health’s review of the genetics White Paper, due in early 2008, to evaluate of the impact of this investment.

**2.7** In 2004 the Human Tissue Act became law. This Act requires that anyone analysing a biological sample to obtain “scientific or medical information about a living or deceased person which may be relevant to any other person (including a future person)” must have appropriate consent from the person from whom the sample was taken for the test to be lawful. This provision specifically addresses concerns raised in *Genes Direct* regarding the potential for non-consensual testing of individuals through home-testing kits.

**2.8** In 2005 the OECD drew up draft *Guidelines for Quality Assurance in Molecular Genetic Testing* after conducting a survey of 18 OECD member countries. The guidelines were adopted by the OECD in May 2007 and contain principles aimed at governments and recommendations for best practice addressed to directors of laboratories, which explain how the principles should be implemented. The OECD believes that genetic testing should be offered through a quality assurance framework that monitors the analytic accuracy of testing and ensures that doctors and patients receive accurate information about a test’s clinical validity and utility. The guidelines also stress the need for ‘truth in advertising’ in any promotional materials and the importance of accurate





and informative test results for clinicians and patients. However, the guidelines address genetic tests for clinical management only; therefore lifestyle tests with no clinical applicability are not covered.

**2.9** In July 2006, the HGC received a response to the *Genes Direct* report from the UK Government (see Appendix 3). This letter thanked the HGC for completing the report and noted several developments in the area. It agreed with the Commission that consumer education had a very important role in minimising the potential harm from direct genetic tests. The Government stated that it was committed to providing ready access to good-quality information about genetic testing for consumers, patients and health professionals. It also warmly supported a proposal for the HGC to host a pan-European meeting on the regulation of direct-to-public genetic tests that would provide a forum for discussion about the development of an effective policy on direct genetic testing services.

**2.10** Also in 2006, the HGC contacted the OFT, the ASA and Ofcom, the independent regulator and competition authority for the UK communications industries, to ask what developments had taken place in relation to the advertising of genetic tests, how they would deal with complaints about genetic tests and whether their respective organisations were co-operating to look into the issue. The HGC also specifically asked Ofcom to what extent it was able to regulate internet content, especially claims made on websites. The responses (see Appendix 4) indicated that the OFT and the ASA work closely together, with the OFT providing the legal support and the ASA being the established route for dealing with complaints. The ASA stated that it would deal with complaints about genetic testing in a similar manner to any complaint, and that their stance on advertising genetic tests is that the advertising claims must be capable of objective substantiation. However, both the ASA and the OFT noted that they would most likely contact the MHRA for further advice when a complaint about genetic testing was received. In response to the query regarding regulation of the internet, Ofcom made it clear that while they were unable to regulate internet content, they acknowledge their role in promoting media literacy and raising consumer awareness of content issues.

**2.11** The HGC has identified a trend in European countries towards limiting the availability of genetic testing direct to the consumer and banning advertising of genetic tests. Some countries, including Switzerland and France, have introduced a universal ban on private genetic testing. However, it is acknowledged that a ban could not prevent the supply of tests over the internet and while those supplying tests could be traced, enforcement is difficult with respect to companies based outside relevant national jurisdictions.

**2.12** The HGC has noted the work of several other groups in this area. EuroGentest is an EU-funded network looking at all aspects of genetic testing with a view to encouraging harmonisation of standards and practice throughout the EU and beyond. It is currently looking at measures to require laboratories and clinical services to be accredited to an International Organization for Standardization (ISO) standard, and is considering the promotion of accreditation for professionals carrying out genetic testing procedures. The Council of Europe is also conducting work reviewing the oversight of genetic tests offered direct to the public, focusing on the issues of consumer education and ensuring that consumers can access information on laboratory accreditation.



**2.13** Despite these developments and the considerable interest, both in Europe and the UK, in reviewing the development of appropriate controls and regulation of genetic tests and genetic testing services, there nevertheless continue to be gaps within which an emerging industry of direct-to-consumer genetic tests is developing without oversight.



## 3. Responding to developments

**3.1** In line with the recommendation in the original 2003 *Genes Direct* report, a follow-up meeting was arranged by the HGC early in 2007 with the aim of reviewing the original recommendations, identifying regulatory gaps and making realistic and practical recommendations for consideration by the HGC, the UK Government and European institutions as appropriate.

**3.2** *Genes Direct* stated the HGC's commitment to liaising with regulators within and outside the EU on the oversight of direct-to-public genetic tests. The meeting was attended by representatives of organisations with key roles in the international oversight of genetic testing, as well as officials from the MHRA and other experts in the field. The meeting also involved members of the HGC's Genetic Services Monitoring Group who have interests in the area and links to both consumer organisations and companies that supply genetic tests. A full list of attendees is supplied at Appendix 1. The HGC would like to record here its gratitude to all those who attended for their willingness to contribute and share their considerable knowledge and expertise.

**3.3** The purpose of the meeting was to advise the HGC on regulatory measures necessary or desirable to protect the public from possible harm arising from genetic testing services supplied directly to the public. The meeting did not seek to return to ground that had already been comprehensively covered in *Genes Direct* but did consider the developments since – and resulting from – that report to identify persistent gaps in the regulatory framework. This was followed by a structured discussion focused on three key areas:

- pre-market review of tests
- quality assurance of testing services
- advertising and promotion, and the provision of independent, impartial advice

**3.4** The full background papers for the meeting are available on the HGC website ([www.hgc.gov.uk](http://www.hgc.gov.uk)). What follows is a summary of the main findings of that meeting and recommendations agreed by the participants and subsequently adopted by the Human Genetics Commission.

### Pre-market review

**3.5** Pre-market review is the assessment and regulation of a test before it is placed on the market. *Genes Direct* recommended that this should include assessment of analytical validity, clinical validity and clinical utility.

**3.6** Participants in the meeting agreed that the scope of pre-market review needs some clarification. Pre-market review typically includes assessment of the claims made by the manufacturer about the product. One view expressed was that pre-market review could be seen as a means of ensuring 'truth in labelling', meaning that the evidence base for all promotional claims, including claims of efficacy, should be assessed. This would set similar standards to those applicable to the marketing of medicines. However, an additional role for pre-market review might be to determine not only the adequacy of the evidence for the test's efficacy but also the appropriateness of marketing the test



directly to the consumer (or certain categories of consumer). Participants therefore agreed that **renewed consideration should be given to the original recommendation in *Genes Direct* that certain genetic tests should only be offered by a suitably qualified health professional.**

**3.7** The IVDD Directive currently covers risk assessment, independent pre-market review and requirements for labelling. However, there is disagreement between EU Member States about the scope of the Directive, i.e. whether it covers all aspects of test performance or is limited to analytical performance of the test. New genetic tests are routinely classified as low risk unless this classification is challenged. There was agreement between participants at the meeting that this default position was not stringent enough. In fact, no new genetic tests have been added to the high-risk list (IVDD Directive, Annex II) since publication of the Directive itself in October 1998. This classification means that no independent evaluation of manufacturers' claims is required before a test comes to market. *Genes Direct* noted the possibility of updating the Annexes of the IVDD Directive and requested that the MDA (now MHRA) continue to seek to ensure that the European Commission works proactively, rather than reactively, to update the Annexes. Participants therefore recommended that **the risk classification of genetic tests covered by the IVDD Directive but currently classified as low risk should be reviewed, as these tests are consequently exempt from the requirement for independent pre-market review.**

**3.8** The IVDD Directive, and consequently the MHRA, is only concerned with devices used for medical purposes. In correspondence, the MHRA has put forward the view that so-called 'lifestyle' tests do not, therefore, fall within their remit. However, this demarcation cannot be relied upon uncritically: if the test manufacturer makes a medical claim for their product, for example that it indicates increased risk of cardiovascular disease, then this may fall within the definition of a medical purpose. Furthermore, it could also be argued that tests which are for the purpose of preventing disease fall within the scope of the Directive, as the EU Directives define medical devices as those intended "for the purpose of diagnosis, prevention, monitoring, treatment or alleviation of disease". They also meet the definition of an IVDD, i.e. their purpose is "providing information concerning a physiological or pathological state". It is noteworthy that the Australian device regulators have recently issued guidance that states that nutrigenetic tests (regarded as a 'lifestyle' test in the UK) will be regulated as IVDDs.

**3.9** In view of the problematic nature of the distinction between 'medical' and 'lifestyle' tests, the participants in the meeting considered the question of whether all genetic tests should fall within the scope of a single regulatory mechanism, irrespective of the IVDD Directive. On that occasion, however, there was agreement that, while consideration should be given to the regulation of genetic tests not covered by the Directive, it would be a mistake to require that all genetic testing products meet the same standards. Participants therefore agreed that **for those tests that fall outside the scope of the IVDD Directive, e.g. 'lifestyle' tests, an alternative regulatory mechanism should be considered to ensure appropriate oversight.**



## Quality assurance

**3.10** The second major regulatory issue of concern to the HGC is quality assurance of a genetic test or service. This involves the various stages in the supply of a genetic test, including issues of availability of information, validation of claims made by the manufacturer, performance of the laboratory carrying out the test, handling of samples and confidentiality. Participants noted that in several relevantly similar areas subject to regulation, codes of practice have been highly effective in maintaining high levels of quality assurance and consumer protection.

**3.11** The draft *OECD Guidelines for Quality Assurance in Molecular Genetic Testing*, adopted by the signatory countries in the summer of 2007, promise to go a long way towards addressing concerns relating to the quality of tests. The guidelines are designed to cover all providers of genetic tests for healthcare purposes and if they are implemented as intended, the principles they set out will guide laboratories in the control of analytical validity through accreditation services and require laboratories to provide data supported by evidence of clinical utility and validity. EuroGentest has also been active in the area of laboratory quality assurance, standards and accreditation. Participants agreed to recommend that **a code of practice relating to genetic testing services should be developed that takes into account the guidelines published by OECD and other relevant international standards, e.g. EuroGentest.**

**3.12** The advantage of such a code of practice, as long as it commands acceptance from all relevant and interested parties, is that it would establish a single, public and transparent benchmark for genetic tests. It would also provide criteria by which consumers could assess the quality of the genetic testing services they are accessing. There are a variety of options for preparing, maintaining and reviewing compliance with such a code and these would require further consideration. To be effective, the development of a code of this kind would, crucially, require the co-operation and commitment of all relevant stakeholders, and participants agreed that **development of a code of practice and its implementation should involve relevant stakeholders including government bodies, public bodies, charities and industry.**

**3.13** One of the criteria of the *OECD Guidelines for Quality Assurance in Molecular Genetic Testing* and of the EuroGentest ISO standard is the requirement for evidence. Since genetic testing generates personal information that must be put in the context of an individual's medical status and background, the OECD guidelines require an evidence base relevant to the individual being tested. A parallel can be drawn with financial services, which, to be appropriate, must take into account the needs of each individual consumer.

**3.14** In *Genes Direct* we concluded that “there is support for considering most genetic tests as if they were ‘prescription-only’”. However, there is no mechanism in place to determine which tests should fall into this category, nor for ensuring that tests are only offered by the appropriate health professional. A recent discussion at the Council of Europe suggests that medical supervision is necessary for the provision of genetic tests depending on an assessment of the ‘seriousness’ of the condition being tested for. Accordingly, a test for a ‘serious’ disease, such as Huntington’s Disease, would need to be offered by a suitably qualified professional. This raises the issue of how to determine which tests should be classified as ‘serious’ and in relation to



seriousness, which healthcare professionals are ‘suitably qualified’ to provide the test. While the question of seriousness in this context is linked to the seriousness of the condition tested for, in our view its focus is more properly located in the possible consequences of the test outcome for the individual concerned. These encompass not only the prognosis but also the individual’s response to the outcome, in terms of significant personal decisions, altered behaviours and psychological effects. Medicinal products are regulated by the European Agency for the Evaluation of Medicinal Products (EMA), which seeks to protect and promote public and animal health through evaluation and supervision of medicines for human and veterinary use. It may be the case that a similar centralised body for diagnostic and prognostic tests for medical purposes would be a useful development. Participants agreed to continue to support the recommendation in *Genes Direct* that specific tests should only be offered through specific outlets or by specific healthcare professionals and that, furthermore, education and training in genetics for healthcare practitioners should be supported; however as this is a recurring theme in many discussions in this area, the specifics of this training were not discussed in detail at the meeting.

### Advice and advertising

**3.15** It was the confirmed view of participants, echoing *Genes Direct* and endorsed following discussion, that **depending on the establishment of a system for classifying genetic tests according to their seriousness, advertisements for tests which it is deemed should only be available via a suitably qualified health professional should be restricted – i.e. no direct-to-public advertising.**

**3.16** On the other hand, a lack of independent consumer information was identified as an important deficit. This relates not only to the provision of information about genetic testing in general, but also to how the evidence used to substantiate claims made about the benefits of tests is assessed. There were concerns that some direct genetic test services made pseudo-health claims, using genetic testing to verify the absence or presence of a particular mutation, but then offering generic health advice or, worse, drawing unsubstantiated health claims from this information. If the consumer is to be able to make a judgement about the validity of claims made by the manufacturer or test provider, then public education is important, supported by transparency and access to the kind of data that is fundamental to the pre-market review process.

**3.17** One suggestion was that test providers should be required to supply a minimum dataset to consumers, with a requirement to place evidence in the public domain. A further step would be for an independent third party to test the evidence for the claims made by manufacturers of all new genetic testing products, whether supplied via a genetic testing service or direct to the consumer.

**3.18** The group agreed that the co-operation of additional bodies was required in order to address the regulation of advertising of genetic tests in the UK. It was suggested that the issue be broached with the ASA and the OFT, as well as the bodies responsible for the content of the advertising codes of practice – the Committee of Advertising Practice (CAP) and the Broadcast Committee of Advertising Practice (BCAP) – to offer to assist in keeping the codes of practice up to date; to share expertise about the sensitive issues involved; and to recommend that consideration be





given to measures that would control the advertising of certain genetic tests through their codes of practice. Claims made on websites are not classed as advertisements for the purposes of regulation. A consensus view that emerged from the meeting was that such claims could be considered equivalent to claims made on instructions for use. It was agreed that the issue of claims made on websites should be also raised with the OFT in relation to the Trade Descriptions Act 1968. Participants agreed that **the HGC should seek to open a dialogue with the ASA, the OFT, the CAP and the BCAP, about enhancing the codes of practice for permitted genetic tests.**

**3.19** Nevertheless, education of consumers was felt to be of paramount importance. It was acknowledged that to accomplish wide-ranging consumer education there would need to be significant involvement from stakeholders, especially in defining categories of tests and the information that should be made available about them. Participants agreed that a system of consumer alerts similar to those used by the US Food and Drug Administration would be useful, but this raised the issue of what body should have the responsibility for issuing them. The HGC itself was suggested as a possible candidate but it was acknowledged that this was not within its current remit.

**3.20** Another acknowledged approach to reducing the risk posed by unreliable information was to encourage the growth of existing public information services offering genetic test summaries. Several independent sources of information (including many websites) were praised as excellent sources of information, although there was concern that they focused mainly on rare genetic diseases and single-gene disorders. A reliance on independent information providers would not therefore ensure comprehensive coverage, and such providers would very likely need to be prompted or offered incentives to deal with the more complex polygenic tests. The Human Variome Project, an international initiative to link all genetic databases, was put forward as a useful model for linking information that is already available, as it included clinical information on genetic conditions. However there are no current plans to make the database available to the public at large. In contrast to this, Lab Tests Online is available to (and intended for) the public, and might be willing to co-operate, if adequately supported, in informing consumers of new tests as they become available. The role of Ofcom in raising consumer awareness could also be encouraged in this area. The group therefore recommended that **the use of existing web-based information sources to provide comprehensive and independent information for consumers should be explored, and test developers/providers should be encouraged to facilitate consumer access to this information.**

**3.21** While it was common ground that comprehensive independent information was required, and that it should be offered through a trusted source that consumers could readily access, there was scepticism about whether the goal of securing, on a website, accurate and up-to-date information on every available genetic test was proportionate and achievable on a voluntary basis. A parallel was drawn with the paternity testing code of practice, whereby websites offering information were listed as part of the code and a role was identified for independent bodies such as the Consumers Association, which has conducted a review of the field in the past and continues to maintain an interest in health screening generally.







## 4. The role of the HGC

**4.1** Participants at the meeting also discussed the part that might be played by the HGC in taking the recommendations forward. As noted above, the suggestion was made that the HGC might take up the regulatory role formerly undertaken by the ACGT. However, the desirability and feasibility of this arrangement were challenged, as the HGC also had a duty to oversee the systems that are put in place and to identify regulatory gaps that have been overlooked. If regulation could be split into three activities: setting standards, information gathering and enforcement, then it was agreed that the HGC should have a role in setting standards and information gathering, but should not be involved in enforcement.





## 5. Consolidated recommendations

**5.1** Below are the recommendations from the original *Genes Direct* report, with the follow-up recommendations from this report inserted where appropriate. As the 2007 meeting focused on the regulatory developments since *Genes Direct*, most of the new recommendations fit into the *Genes Direct* recommendations relating to regulation.

**5.2** **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** (*Genes Direct*, paragraph 3.24). This should not mean that people face difficulty accessing appropriate genetic testing or health information about themselves.

**5.3** **We feel strongly that there should be a well-funded NHS genetics service supported by a genetically literate primary care workforce, which can properly manage and allow access to new predictive genetic tests that are being developed** (*Genes Direct*, paragraph 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.

**5.4** **In view of this, we conclude that most genetic tests that provide predictive health information should not be offered as direct genetic tests** (*Genes Direct*, paragraph 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 although some may be provided via pharmacists and others, if they are low risk, can be bought in any shop.

**5.5** 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.

**5.6** **Renewed consideration should be given to the original recommendation in *Genes Direct* that certain genetic tests should only be offered by a suitably qualified health professional** (paragraph 3.6 of this report).

**5.7** **We have concerns about predictive genetic tests that are done at home** ('direct-to-consumer') (*Genes Direct*, 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.

**5.8** **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** (*Genes Direct*, paragraph 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 (*Genes Direct*, paragraph 3.40).

- **The risk classification of genetic tests covered by the IVDD Directive but currently classified as low risk should be reviewed, as these tests are consequently exempt from the requirement for independent pre-market review** (paragraph 3.7 of this report).
- **For those tests that fall outside the scope of the IVDD Directive, e.g. ‘lifestyle’ tests, an alternative regulatory mechanism should be considered to ensure appropriate oversight** (paragraph 3.9 of this report).
- **A code of practice relating to genetic testing services should be developed that takes into account the guidelines published by OECD and other relevant international standards, e.g. EuroGentest** (paragraph 3.11 of this report)
- **Development of a code of practice and its implementation should involve relevant stakeholders including government bodies, public bodies, charities and industry** (paragraph 3.12 of this report).
- We welcome the proposed arrangements for reviewing genetic tests, which will be introduced by the **UK Genetic Testing Network (UKGTN)** of the Genetics Commissioning Advisory Group (GenCAG). We believe that this work may provide useful basis for the oversight of direct genetic tests (*Genes Direct*, paragraph 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 (*Genes Direct*, paragraph 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 (*Genes Direct*, paragraph 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 Healthcare Professionals** [now called the Council for Healthcare Regulatory Excellence] 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 (*Genes Direct*, paragraph 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 emphasise the need for responsible and accurate advertising of such products (*Genes Direct*, paragraph 3.59).
- **Depending on the establishment of a system for classifying genetic tests according to their seriousness, advertisements for tests which it is deemed should only be available via a suitably qualified health professional should be restricted – i.e. no direct-to-public advertising** (paragraph 3.15 of this report).
- **The HGC should seek to open a dialogue with the ASA, the OFT, the CAP and the BCAP, about enhancing the codes of practice for permitted genetic tests** (paragraph 3.18 of this report).
- 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 (*Genes Direct*, paragraph 3.62).
- **The use of existing web-based information sources to provide comprehensive and independent information for consumers should be explored, and test developers/providers should be encouraged to facilitate consumer access to this information** (paragraph 3.20 of this report).
- 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 (*Genes Direct*, paragraph 3.63).



## Glossary

ACGT	Advisory Committee on Genetic Testing
Analytical validity	accuracy of test in identifying the biomarker
ASA	Advertising Standards Authority
BCAP	Broadcast Committee of Advertising Practice
CAP	Committee of Advertising Practice
Clinical utility	likelihood that test will lead to an improved outcome
Clinical validity	relationship between the biomarker and clinical status
CPA	Clinical Pathology Accreditation
DTC	direct-to-consumer – where the test is provided without an intermediary between the consumer and the test provider
DTP	direct-to-public – where the test is provided via a non-medical intermediary, such as a pharmacist or alternative health practitioner
FDA	US Food and Drug Administration
GenGAG	Genetics Commissioning Advisory Group
HGC	Human Genetics Commission
ISO	International Organization for Standardization
IVDD Directive	EU In Vitro Diagnostic Devices Directive
MHRA	Medicines and Healthcare products Regulatory Agency
NHS	National Health Service
OECD	Organisation for Economic Co-operation and Development
Ofcom	Office of Communications
OFT	Office of Fair Trading
UKGTN	United Kingdom Genetic Testing Network





## Appendix 1 – Meeting participants

**Mr Geoffrey Watts (Facilitator)**

Presenter, BBC Radio programme  
'Leading Edge'

**Dr Mark Bale**

Head of Genetics Branch,  
Scientific Development & Bioethics  
Division,  
Department of Health, UK

**Professor Jean-Jacques Cassiman**

Co-ordinator  
EuroGentest

**Dr Robert Elles**

Director of National Genetics Reference  
Laboratory, Manchester

**Professor Peter Furness**

Chair of Specialist Advisory Committee  
in Histopathology,  
Royal College of Pathologists

**Dr Courtney Harper**

Office of In Vitro Diagnostic Device  
Evaluation and Safety, FDA

**Dr Chris Hodges**

Associate Fellow at the Centre for  
Socio-Legal Studies,  
Oxford University

**Mr Stuart Hogarth**

Research Associate in the Epidemiology  
for Policy Group,  
Department of Public Health and  
Primary Care,  
University of Cambridge

**Ms Gail Javitt**

Adjunct Professor of Law,  
Genetics and Public Policy Center,  
Johns Hopkins University

**Dr Mark Kroese**

Consultant in Public Health Medicine,  
Public Health Genetics Unit [now the  
PHG Foundation]

**Dr Susanne Ludgate**

Director of Clinical Devices Division,  
MHRA

**Mme Laurence Lwoff**

Bioethics Department,  
Council of Europe

**Dr Elettra Ronchi**

Biotechnology Unit,  
Directorate for Science,  
Technology and Industry,  
OECD

**Dr Paula Saukko**

Economic & Social Research Council  
Centre for Genomics in Society,  
University of Exeter

### HGC

**Dr Celia Brazell**

HGC Commissioner

**Miss Sarah Connelly**

HGC Secretariat

**Dr Paul Debenham**

HGC Commissioner

**Dr Iona Heath**

HGC Commissioner (to July 2007)

**Mr Alastair Kent**

HGC Commissioner

**Mr Chris Lucas**

HGC Secretariat

**Dr Christine Patch**

HGC Commissioner

**Mr Richard Pitts**

Office of Science and Innovation

**Peter Sayers**

HGC Commissioner



## Appendix 2 – Companies offering genetic testing services in the UK

### UK-based companies offering DTC and/or DTP tests

Company	Test(s) offered	Delivery model
Genetic Health	<p>Female Plus – genetic predisposition to breast cancer, osteoporosis, thrombosis, cancer and long-term exposure to oestrogens</p> <p>Male Plus – genetic predisposition to prostate cancer, thrombosis, osteoporosis, metabolic imbalances of detoxification and chronic inflammation</p> <p>Nutrigenetic test – test for a range of genes which influence nutritional processes such as lipid and glucose metabolism</p> <p>Pharmacogenetic – test for CYP450 genes, which influence how the liver metabolises a large number of commonly prescribed drugs</p> <p>Premium Male Gene/Premium Female Gene – combine all the other tests except the nutrigenetic one</p> <p>(Tests are performed by Austrian test developer and laboratory Genosense)</p>	DTC via internet; Plus and Premium services include a medical consultation
G-nostics	NicoTest – test for predisposition to nicotine addiction and response to nicotine replacement products	DTC via internet and also DTP through pharmacies
Medi-Checks	Wide range of well-established genetic tests, from Factor V thrombosis risk to BRCA testing for breast cancer risk (tests are performed by the private pathology laboratory TDL)	DTC via internet but company recommends physician referral for high-impact tests such as BRCA

Internet access means that UK consumers can order tests from providers outside the UK. As the country with the broadest range of providers, it is instructive to note the range of DTC/DTP companies in the US, as well as those operating in Europe.

### European companies offering DTC and/or DTP tests

Company	Test(s)	Delivery model
Geneticom (Netherlands)	Common disease risk	Not clear
Genosense (Austria)	Susceptibility tests	Do not offer DTC themselves but some of their ‘partner physicians’ in other countries seem to offer DTC, e.g. Genetic Health in UK
Medigenomix (Germany)	Thrombophilia and osteoporosis risk	DTC via internet

### Non-European-based companies offering DTC and/or DTP tests

Company	Test(s) offered	Delivery model
23andMe (US)	Company offers susceptibility testing for common diseases as well as ancestry testing	DTC via internet
Acu-Gen Biolab, Inc. (US)	Fetal DNA gender test	Blood spot obtained by consumer using materials provided by company
Consumer Genetics (US)	Alcohol metabolism Asthma drug response Caffeine metabolism Fetal gender	DTC via internet
Cygene Direct (US)	Athletic performance Glaucoma and Macular Degeneration Osteoporosis Thrombosis	DTC via internet

Company	Test(s) offered	Delivery model
DeCODE (Iceland)	deCODEme test for genetic variations associated with 17 common conditions: Age-related macular degeneration, Asthma, Atrial fibrillation, Breast Cancer, Celiac Disease, Colorectal Cancer, Exfoliation Glaucoma XFG, Inflammatory Bowel Disease, Multiple sclerosis, Myocardial Infarction, Obesity, Prostate cancer, Psoriasis, Restless legs, Rheumatoid arthritis, Type 1 Diabetes and Type 2 Diabetes	DTC via internet
Dermagenetics (US)	Dermagenetics skin DNA profile DNA UltraCustom skin cream	DTC through spas and similar retailers
DNADirect (US)	Alpha-1 Antitrypsin deficiency Ashkenazi Jewish carrier screening Blood clotting disorders Breast and ovarian cancer Colon cancer screening (PreGen-Plus) Cystic fibrosis Diabetes risk (deCODE T2™) Drug response panel Haemochromatosis Infertility Recurrent pregnancy loss Tamoxifen	DTC via internet  Personalized report provided online  Genetic counsellors available by phone
Genelex (US)	PGx Testing Coeliac disease DNA Diet™ consultation Gum disease Haemochromatosis Nutritional genetic testing Rapid Results DNA Diet Weight Loss System	DTC via internet



Company	Test(s) offered	Delivery model
Graceful Earth (US)	Alzheimer (ApoE)	DTC via internet
Health Tests Direct (US)	More than 400 blood tests listed on site, including a few genetic tests (cystic fibrosis carrier screen, Factor V Leiden)  Others may also be available by calling	DTC via internet
HealthCheckUSA (US)	A wide range of laboratory tests including the following genetic tests:  Coeliac disease  Factor V Leiden  Factor V R2  Hereditary haemochromatosis	DTC via internet  As additional service, patient can request interpretation by board-certified physician. Free genetic counselling offered by Kimball Genetics for physicians, patients, and families
Holistic Heal (US)	Nutrigenomic test: comprehensive methylation panel with methylation pathway analysis  Company also sells a variety of nutritional supplements	Not described
Kimball (US)	Wide range of well-established genetic tests	DTC via internet but detailed telephone consultation with certified genetic counsellor is mandatory. Report is sent to physician and customer

Company	Test(s) offered	Delivery model
Mygenome.com (US)	<p>Alzheimer's disease – genetic testing for common risk factors</p> <p>Cardiovascular disease – genetic tests differentiate treatable risk factors for heart disease and stroke</p> <p>Drug sensitivities – genetic tests for genes that affect the safety and activity of many common prescription and over-the-counter drugs</p> <p>Osteoporosis – genetic tests identify risk factors for osteoporosis and fractures</p> <p>Pregnancy risk – genetic tests identify risk factors for complications of pregnancy</p> <p>Thrombosis – genetic tests identify risk factors for blood clots</p>	Not clear
Navigenics (US)	No tests on market yet but company will offer whole-genome scanning and risk analysis for more than 20 common diseases such as prostate cancer and diabetes	No tests offered yet
Quixtar (US)	<p>Gensona General Nutrition Genetic Test</p> <p>Gensona Heart Health Genetic Test (IL1 gene)</p> <p>Nutrigenomic Dietary Supplement</p> <p>Nutrilite® IL1 Heart Health</p>	DTC via internet.
Salugen (US)	<p>Genoscore DNA Test (nutrigenetic)</p> <p>GenoTrim™ Nutrigenomic Supplement</p> <p>SpaGen™ supplements</p>	<p>Not clear</p> <p>SpaGen sold through spas</p>
Sciona (US)	<p>Antioxidant/detoxification</p> <p>Bone Health</p> <p>Heart Health</p> <p>Inflammation health</p> <p>Insulin resistance</p>	DTC via internet

Company	Test(s) offered	Delivery model
Smart Genetics (US)	HIVmirror™ (test to predict how quickly HIV may become AIDS using the CCR5-Delta 32 and CCR2-64I gene)	DTC via internet, free counselling available
Suracell (US)	Core nutrition: essential genetic formula  Personal DNA Analysis Profile test – DNA profile test that identifies your inherited genetic ageing profile, and a biomarker assessment test that measures DNA damage, oxidative stress and free radical levels  Repair: personal genetic formulations	DNA sample is obtained from a “simple mouthwash rinse”  Urine sample “provides biomarkers for the assessment test”  Optional lifestyle questionnaire can be completed

The information contained in this appendix was up to date on 27 November 2007. In compiling this appendix, the HGC is grateful for the assistance of Stuart Hogarth of the University of Cambridge University and, for the US data, the Genetics and Public Policy Center.

### Types of offer

While there are no hard figures to illustrate the size of the genetic testing market, it is clear that the delivery of genetic tests in the UK continues to be overwhelmingly dominated by NHS provision. Unlike the US, there is relatively little commercial provision and, even among commercial providers, there is only limited DTC/DTP provision. It is unclear how large a market there may be for such services in the future, but the pathology modernisation process within the NHS is likely to result in a greater role for commercial clinical laboratories and there has been a slow but steady emergence of commercial test providers. Furthermore, the public can use the internet to access testing services outside the UK.

As is clear from the above listings, the range of delivery models varies considerably. Some testing services are delivered, primarily, directly to the consumer via the internet; others rely more heavily on direct-to-public provision. The level of service offered varies, with some companies requiring pre- and post-test counselling as a mandatory part of the testing process, others offering such services as an optional extra for which there is an additional fee. There is a similar variation in the type of tests offered, ranging from tests which are a well-established part of clinical genetics, through to more dubious susceptibility and nutrigenetic tests for which there is very little clinical evidence.

Another business model has emerged involving a third party. Companies, such as DNA Direct in the United States, which are neither a test manufacturer nor a reference laboratory but who offer a range of genetic tests directly to consumers are, in effect,



intermediaries between reference laboratories and doctors and patients. Similar arrangements can be seen in the UK, for instance the company Medi-Checks, which offers a wide range of tests directly to the consumer via the internet in collaboration with the private pathology laboratory TDL. Genetic Health are a London-based company, which offers a range of tests developed and performed by the Austrian company Genosense. Genosense has partners across the globe.

These companies operate within a regulatory gap because they are neither conventional device manufacturers nor pathology laboratories, and may therefore not fall under either the regulation governing medical devices or the quality assurance framework for laboratories. It is possible to regulate the laboratory which develops and provides the tests, but not the intermediary. Thus a company that has developed a new test and wants to make strong clinical claims for their test directly to consumers without regulatory scrutiny of those claims could do so via such an intermediary.



## Appendix 3 – UK Government response to the HGC's *Genes Direct* report

*From The Minister of State  
Andy Burnham MP*

MS(DQ)50228

Baroness Helena Kennedy QC  
Human Genetics Commission  
605 Wellington House  
133-155 Waterloo Road  
London  
SE1 8UG

10th July 2006

Dear Helena,

### **Human Genetics Commission: Oversight of genetic tests supplied directly to the public**

Thank you for your letter to Jane Kennedy of 31 March 2006 about the Human Genetics Commission's report *Genes Direct* and the oversight of genetic tests that are supplied directly to the public. I am replying as the new health minister with responsibility for genetic issues. This letter contains a joint view from the Department of Health and Lord Sainsbury, the Science Minister.

At the time of *Genes Direct's* publication, the Government welcomed the advice that the HGC gave in the report and recognised the need to find a balance between the right of individuals to have information about their own health and the need to protect vulnerable groups, particularly children. I would like to take this opportunity to set out some developments that have been taken forward since the publication of the report. *Genes Direct* recognised the need for a well-funded NHS genetics service and the genetics White Paper, published in May 2003, set out an additional investment of £50 million in genetics in England. I am also aware that the devolved administrations have been making similar investments in this area. This extra investment in funding and training has gone a long way towards meeting the HGC's main recommendation that the main route that people want to access predictive genetic testing is via their own GP or primary healthcare team and possibly referral to NHS specialists. This is the route where they can get the counselling that they need and appropriate consent can be obtained for proven and high quality genetic tests.

The Human Tissue Act, which comes into force later this year, helps place a responsibility on companies that provide private DNA testing to ensure that appropriate measures are in place to ensure that DNA testing is done with full and lawful consent. This will address another of your recommendations against the provision of services which may encourage non-consensual testing.

The Government agrees with the Commission that consumer education plays an important role in minimising the potential harms that may follow from direct genetic



tests. We are committed to providing ready access to good quality information about genetic testing, for consumers, for patients and for health professionals and the White Paper provided investment aimed at developing information on all aspects of genetic testing and advances in genetic knowledge.

The Government remains grateful to the HGC for its helpful and constructive comments on possible regulatory mechanisms for this area. I am particularly grateful for your pragmatic suggestion of how to take this work forward now that it is clear that the MHRA is not able – under UK and EU law – to operate in the areas that you advocated. I warmly support your proposal that HGC should host a pan-European meeting on the regulation of direct-to-public genetic tests. HGC should fund this event out of its existing budget. Such an event could provide useful opportunities to consider work being undertaken by international partners, for example including the Council of Europe’s Steering Committee on Bioethics.

While the commercial market for genetic testing in this area is still in its relative infancy this seems like an opportune moment to have this discussion about appropriate standards and controls. This forum will provide a valuable focus for the development of an effective policy on direct genetic testing services. Many of the issues that you raised in *Genes Direct* in 2003 are still current and the report continues to provide a useful framework for further discussion in this area.

I am copying this response to Health Ministers in Scotland, Wales and Northern Ireland.

I would like to take this opportunity to say that I look forward to working with you and the Human Genetics Commission as the new minister at the Department of Health.

Yours sincerely,

Andy Burnham

## Appendix 4 – Correspondence regarding the advertising of genetic tests

### Letter from HGC to the Content Board of Ofcom

Content Board  
OFCOM  
Riverside House  
2a Southwark Bridge Road  
London  
SE1 9HA

28 March 2006

Dear Sir/Madam,

I am writing to you as the Secretary of the Human Genetics Commission (HGC), the UK Government's advisory body on human genetics.

Following a request from Government in 2002, the HGC conducted a review of genetic testing services supplied direct to the public. Their findings were published in 2003 in their report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. I enclose a copy for your information.

Since publication of the report, the Commission has continued to monitor progress in this area and although this market has not expanded in the way that was envisaged, many more direct-to-public genetic tests are now available over the internet. The Commission is very concerned about this development as several of these internet companies provide a service that provides genetic tests with little counselling or medical intervention. In addition, as some samples for testing are collected by the consumer and returned by post, this leads to a greater risk that these tests are performed without appropriate and legally valid consent being gained, especially when children are involved.

With this in mind, I would be grateful if you would be able to provide the Commission with information about the role of OFCOM and the Content Board with regards to internet content. I would also be interested to hear about the steps that OFCOM is taking to make the public aware that content on the internet is not subject to the same regulation as other areas. Specific issues that it would be very helpful to have information on include:

- Is the issue of providing genetic testing services being monitored by OFCOM?
- Is monitoring of content that has not been triggered by consumer complaint an activity that OFCOM undertakes?
- If an internet complaint is brought to OFCOM's attention, how do you handle it, given your statutory responsibilities?

In addition, the *Genes Direct* report identified a key role for the Advertising Standards Authority and the Office of Fair Trading. How would OFCOM work with these bodies in future cases where issues might cover several remits?



I would be very grateful if you could share any information that you can with me on this issue. Please let me know if you require any further information.

Yours faithfully,

Gwen Nightingale  
Secretary to the Human Genetics Commission

## Response from Ofcom to HGC

12 April 2006

Gwen Nightingale  
Secretary to the Human Genetics Commission  
6th Floor North  
Wellington House  
133-155 Waterloo Road  
London  
SE1 8UG

Dear Ms Nightingale

Thank you for your letter of 28th March to the Ofcom Content Board regarding your concern about the genetic tests available over the internet and enquiring about the role of Ofcom and the Content Board with regards to internet content.

The Communications Act does not give Ofcom a role in the regulation of content over the internet. However, Section 11 of the Communications Act gives Ofcom a duty to promote media literacy.

Specifically it requires Ofcom to take such steps, and to enter into such arrangements, as appear to them calculated to bring about, or to encourage others to bring about, a better public understanding of the nature and characteristics of material published by means of the electronic media. Publication includes by an electronic communications network to members of the public or of a section of the public. In this respect Ofcom's role is to raise awareness of the nature of the content rather than regulate it.

In the discharge of this duty Ofcom seeks partnership with others. We are working closely with the internet sector to raise awareness and provide tools for people to manage their online experience. We are members of the Home Office Task Force on internet safety which has over the last few years produced a number of 'best practice' guides to ISP. Details can be found on the Home Office website at:

<http://police.homeoffice.gov.uk/operational-policing/crime-disorder/child-protection-taskforce>

We are active supporters of the UK Awareness Node of the European Safer Internet Action Plan. Details of this group can be found at:

[http://www.uclan.ac.uk/host/cru/isca\\_overview.htm](http://www.uclan.ac.uk/host/cru/isca_overview.htm)

We work closely with a number of other organisations to raise awareness of issues related to internet content and services. I would be happy to provide further information if this would be helpful. Genetic testing services have not been raised as a concern by these organisations and I'm grateful for you bringing it to our attention.

The regulation of broadcast advertising content has been contracted out to the ASA by Ofcom. The ASA therefore now regulates all broadcast and non-broadcast advertising content. Ofcom does not get involved in any of the ASA's casework or policy issues, unless required by the ASA.



Ofcom has no formal relationship with the OFT in relation to broadcast content issues, except via the Control of Misleading Advertisements Regulation, when Ofcom is designated as a competent body.

Best wishes

Tim Suter



## Letter from HGC to ASA

Advertising Standards Authority  
Mid City Place  
71 High Holborn  
London  
WC1V 6QT

28 March 2006

Dear Sir/Madam,

I am writing to you as the Secretary of the Human Genetics Commission (HGC), the UK Government's advisory body on human genetics.

Following a request from Government in 2002, the HGC conducted a review of genetic testing services supplied direct to the public. Their findings were published in 2003 in their report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. One of the recommendations of the report was that the ASA should emphasise the need for responsible and accurate advertising of such products. I enclose a copy of the report for your information.

Since publication of *Genes Direct*, the Commission has continued to monitor progress in this area and although this market has not expanded in the way that was envisaged, many more direct-to-public genetic tests are now available over the internet. The Commission is very concerned about this development as several of these internet companies provide a service that provides genetic tests with little counselling or medical intervention. In addition, as some samples for testing are collected by the consumer and returned by post, this leads to a greater risk that these tests are performed without appropriate and legally valid consent being gained, especially when children are involved.

You may recall that the ASA took action in relation to misleading adverts for "Genetic Hair" in 2003. At this time you indicated that you were keen to engage HGC for advice on specialist aspects of your work evaluating complaints relating to genetics. The Commission would be interested to hear whether there have been any other developments since this complaint? They would also be interested to hear how the ASA would deal with similar complaints about genetic products in future, especially when other regulatory bodies might also be involved, for example OFCOM and the Office of Fair Trading. How would the ASA work with these bodies in future cases where issues might cover several remits?

I would be very grateful if you could share any information that you can with me on this issue. Please let me know if you require any further information.

I have sent a similar letter to OFCOM and the Office of Fair Trading.

Yours faithfully,

Gwen Nightingale  
Secretary to the Human Genetics Commission



## Response from ASA to HGC

Ms G Nightingale  
Human Genetics Commission  
6th Floor North, Wellington House  
133-155 Waterloo Road  
London SE1 8UG

21 April 2006

Dear Ms Nightingale,

Thank you for your letter of 28 March 2006 in which you enquired about developments in the Advertising Standards Authority's (ASA) policy towards advertisements for genetic testing services.

The ASA is responsible for ensuring that all advertisements, wherever they appear, are legal, decent, honest and truthful.

Since your complaint and the publication of your report in 2003, we have not received any further complaints about advertisements for genetic testing kits or for products claiming to use genetic technology. This seems to reinforce your point that the market has not expanded as envisaged.

Therefore, our position on advertising for such services is the same as that which we adopt towards all advertising: that advertising claims must be capable of objective substantiation, regardless of the product or service being advertised. If we were to receive a complaint about an ad for a genetic testing service, it would be assessed against the relevant clauses of the Codes relating to misleadingness, health claims, or responsibility, as appropriate. For your reference I have enclosed a copy of our original response to your consultation, which includes further details of the specific Code clauses that might be used in these cases.

If we were to receive a complaint about a genetic testing service ad, it would first be assessed to see whether the ad fell within the scope of the Advertising Codes. Your letter mentioned that you were specifically concerned that many of these products were marketed on the internet. The remit of the Code for online advertising is restricted to ads in "paid for" space, e.g. banner and pop-up advertisements, ads in commercial e-mails and sales promotions (wherever they appear online, including in organisations' websites or e-mails). The Code does not apply to organisations' claims on, and the content of, their own websites. This means that we would advise complainants about this type of content to raise their concerns with their local Trading Standards Office. This course of action would also be followed for any other complaints that fell outside the scope of the Code, but which might be considered by Trading Standards.

If the advertised product was a testing kit that offered to treat or diagnose an adverse medical condition, it would be subject to statutory control under the Medicines Act 1968. Therefore, complaints in this category would be directed to the Medicines and Healthcare Products Regulatory Agency for investigation.

Your letter also enquired about our relationship with other regulators, including Ofcom and the Office of Fair Trading (OFT). In November 2004, Ofcom contracted-out the regulation of TV and radio advertising to the ASA. The ASA is now the “one-stop-shop” for advertising complaints in the UK. Therefore, any complaint received about a genetic testing service advertised on radio or television would fall within our remit. We have not received any complaints about TV or radio advertising for these kinds of products since November 2004.

We also have a close working relationship with the OFT. The ASA is regarded as the ‘established means’ for dealing with misleading non-broadcast ads under the Control of Misleading Advertisements Regulations 1988 (as amended), with the OFT operating as our legal backstop for the purposes of this legislation. We have established Memorandums of Understanding with both Ofcom and the OFT, which set out agreed procedures of operation and case-handling principles.

Our contract with Ofcom means that we are required to handle all complaints about TV and radio advertising. However, when investigating complaints about ads in non-broadcast media that fall within another regulator’s remit, the ASA’s normal practice is to liaise with the other regulator in the first instance to decide which is the most appropriate body to take action. If the issue being raised comes under statutory control, then it is likely that we would refer the matter to the relevant statutory regulator. If the ASA decided to investigate the complaint, we would keep the other relevant regulators informed throughout the case. Of course, if we were to receive any complaints about genetic testing services, we would be keen to liaise with the HGC for specialist advice.

I do hope that this letter has been helpful in answering your queries. Please do not hesitate to contact me if you require any further information about this matter or, indeed, if you have any questions about the work of the ASA. Alternatively you can visit our website [www.asa.org.uk](http://www.asa.org.uk).

Yours sincerely

Lynsay Taffe  
Policy & Public Affairs Advisor



## Letter from HGC to OFT

Office of Fair Trading  
Fleetbank House  
2-6 Salisbury Square  
London EC4Y 8JX

28 March 2006

Dear Sir/Madam,

I am writing to you as the Secretary of the Human Genetics Commission (HGC), the UK Government's advisory body on human genetics.

Following a request from Government in 2002, the HGC conducted a review of genetic testing services supplied direct to the public. Their findings were published in 2003 in their report *Genes Direct: Ensuring the effective oversight of genetic tests supplied directly to the public*. I enclose a copy of this report for your information. One of its recommendations was that the OFT, along with the Advertising Standards Authority, should emphasise the need for responsible and accurate advertising of such products. The Commission also suggested that an OFT endorsed self-regulatory code of practice in this area might be useful, but to date this has not been taken forward.

Since publication of *Genes Direct*, the Commission has continued to monitor progress in this area and although this market has not expanded in the way that was envisaged, many more direct-to-public genetic tests are now available over the internet. The Commission is very concerned about this development as several of these internet companies provide a service that provides genetic tests with little counselling or medical intervention. In addition, as some samples for testing are collected by the consumer and returned by post, this leads to a greater risk that these tests are performed without appropriate and legally valid consent being gained, especially when children are involved.

The Office of Fair Trading has a strong role to play in promoting and protecting consumer interests throughout the UK and the Commission would be interested to hear how you might get involved when a company was behaving irresponsibly towards consumers in this area. The Commission would also be interested to hear about any feedback you have received from your excellent misleading adverts campaign.

I would be very grateful if you could share any information that you can with me on this issue. Please let me know if you require any further information.

Yours faithfully,

Gwen Nightingale  
Secretary to the Human Genetics Commission

## Response from OFT to HGC

Ms Gwen Nightingale  
Secretary to the Human Genetics  
Commission  
6th Floor North  
Wellington House  
133-155 Waterloo Road  
London SE1 8UG

Date 10 April 2006

Dear Ms Nightingale,

Thank you for your letter of 28 March concerning OFT's possible interest in genetic testing services.

Firstly, it may be helpful if I explain the OFT's powers in relation to misleading advertising.

Under the Control of Misleading Advertisements Regulations 1988 ("CMARs") the OFT can bring civil proceedings for a court injunction to stop a misleading claim where no undertaking is given to us to amend or discontinue it. Where an advertiser breaches a court injunction, we can invite the court to punish them for contempt which could result in a fine or imprisonment. For the purposes of the CMARs, an advertisement, including internet advertising, is misleading if in any way it deceives, or is likely to deceive, the persons to whom it is addressed and if, by reason of its deceptive nature, it is likely to affect their economic behaviour.

However, the CMARs also encourage the OFT to give organisations, which already exist, such as the Advertising Standards Authority, to control advertising, the opportunity to deal with complaints in the first instance. In this connection, the OFT would consider the Medicines and Healthcare products Regulatory Agency ("MHRA"), to be best placed to advise and take action in relation to complaints about misleading advertising claims for medical devices such as genetic testing devices/services.

In addition to the above, the OFT has the power under Part 8 of the Enterprise Act 2002 to apply for an injunction against a business which has engaged in conduct which infringes specified consumer protection enactments, or rules of law, and harms the collective interests of consumers.

On the basis of the information provided, it is not clear that the points you have raised about the ethical and legal use of genetic testing devices would fall to be considered by the enactments specified for the purposes of Part 8 of the Enterprise Act 2002.



However, the OFT remains open to further discussions on the potential of an OFT approved code within this area. If there are points you would like to explore please contact Steve Hill, Head of Mis-selling of Goods and Services at the above address or at [steve.hill@oft.gsi.gov.uk](mailto:steve.hill@oft.gsi.gov.uk).

I hope that this is helpful.

Yours sincerely

Mrs Christine Wade MBE  
Director, Consumer Regulation Enforcement Division





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