A Common Framework of Principles for direct-to-consumer genetic testing services

Principles and Consultation Questions

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Introduction

The Human Genetics Commission is seeking views on a Common Framework of Principles for direct-to-consumer genetic testing services.

The direct-to-consumer genetic testing market has grown steadily over the last 10 years. The majority of test providers are located in the United States and Europe; however, tests are marketed over the internet, making services readily available to consumers worldwide. In the majority of countries, there is very little regulation of this rapidly expanding market. Members of the public are therefore able to purchase a wide range of both health and recreational genetic tests where the quality of the service and the provision of support to accompany testing is determined by the test provider. Yet a genetic test result, regardless of whether it is considered to be health related or recreational, can significantly influence choices that may profoundly affect the life of an individual and their family. Genetic tests that can provide information about health can heighten anxieties, leading individuals to make inappropriate requests for further tests, or alternatively encourage a complacent disregard for the effects of an unhealthy lifestyle. Other tests can also provoke anxieties, such as ancestry tests that offer fundamental information about identity and ethnicity. Recognising this, it is encouraging that many organisations and individuals, including the direct genetic test providers themselves, support the development of an international set of Principles.

Purpose and scope of the Principles

The Common Framework of Principles will promote high standards and consistency in the provision of direct-to-consumer genetic tests amongst commercial providers at an international level in order to protect the interests of people seeking genetic tests and their families. They will identify where individual companies and or national jurisdictions should have defined measures in place and the nature of those measures. These Principles cover all aspects of direct-to-consumer genetic testing services, including the marketing and advertising of services, the collection, analysis and storage of biological samples, the interpretation of results and the provision of results to the consumer. They are applicable to the provision of all genetic tests, when it is possible for a consumer to purchase a test without prescription by a qualified medical professional subject to statutory regulation, and in situations where information about the availability of tests is targeted at consumers rather than qualified medical professionals. This includes all tests that provide health information, carrier tests, pharmacogenetic tests, lifestyle or behavioural tests, nutrigenetic tests, phenotype tests, genetic relatedness tests, ancestry tests and tests to determine genetic matches.

The UK Human Genetics Commission

The Human Genetics Commission is the UK Government's advisory body on developments in human genetics and how they impact on individuals and society. It advises Ministers in the UK on the potential ethical and legal implications of human genetic knowledge and its applications for health and economic and social well-being.

The HGC has published two internationally respected reports on direct-to-consumer genetic testing services, Genes Direct (2003) and More Genes Direct (2007). It is well placed to lead the development of a Common Framework of Principles for direct-to-consumer genetic tests. It maintains contacts with equivalent bodies in other countries.
The HGC is not a regulatory body. It hopes that these Principles will lead to the development of codes of practice that take account of existing regulatory structures where the need for additional regulation or legislation is revealed to be necessary.

The development of the Principles

The need to develop high level Principles for direct genetic tests was first identified during a workshop, convened by the HGC, to discuss the desire for a UK code of practice. Whilst this workshop identified an appetite for specific guidance on good practice, a key outcome was the recognition that the provision of direct genetic testing services took place in an international market that crossed national borders and regulatory jurisdictions. In order to promote consistency the decision was made to initiate a Common Framework of Principles for direct-to-consumer genetic testing services as a high level document having general applicability across all jurisdictions. The Principles were developed by a collaborative working group that was convened and supported by the Human Genetics Commission. The expertise of the working group includes the direct genetic testing industry, clinical and molecular geneticists, genetic counsellors, experts in regulation and those with experience in offering support to individuals with genetic conditions. The development of consultation questions was guided by issues raised by members of the expert working group and by HGC Commissioners. Whilst there has been an overall consensus amongst working group members and Commissioners about the content of the Principles, on occasions views have differed on the emphasis that should be placed on a specific point. This is highlighted in the consultation questions that follow. Responses from this consultation will help to identify the most broadly acceptable solutions.

We welcome comments on these principles in any form and we would be especially grateful for considered responses to any or all of the questions that follow. We hope to elicit views from the broadest range of respondents possible.

How to respond to the consultation

We have set out below 11 questions. It would be especially helpful if respondents could indicate the reasons for their responses including any arguments or evidence if appropriate.

A Common Framework of Principles for direct-to-consumer genetic testing services

These Principles have been developed by a collaborative working group comprising representatives from the genetic testing industry, experts in regulation, clinical and molecular genetics and genetic counselling, representatives from groups that support individuals with genetic conditions and the UK Department of Health.

The group was convened and supported by the UK Human Genetics Commission, the UK Government's advisory body on developments in human genetics and their ethical, legal, social and economic implications.

Purpose

The purpose of these Principles is to promote high standards and consistency in the provision of genetic tests amongst commercial providers at an international level in order to safeguard the interests of people seeking genetic testing and their families. They identify areas where individual providers, professional organisations, regulatory bodies, and/or national jurisdictions should have defined measures in place, and the nature of those measures. The Principles should be used in accordance with applicable international instruments and domestic law.

Genetic test results have the ability to give rise to a broad spectrum of responses. How an individual responds to the results of a genetic test will depend, sometimes to a great extent, on the personality of that individual and their understanding of the test result and its implications. Therefore, the consequences for an individual of taking a genetic test will vary significantly depending on the individual taking a test, the information provided and the context in which the test is taken. When a genetic test is provided outside a framework of healthcare, special attention must be given as to how that individual may respond to the results of the genetic test and the subsequent impact the test results may have on that individual and their family. With this in mind, these Principles have been developed with the best interests of consumers at the forefront.

Scope

These Principles are intended to cover all situations in which it is possible for a private consumer to purchase a genetic test without prescription by a qualified medical professional subject to statutory regulation. The Principles cover:

- tests that are provided directly to the public without an intermediary between the consumer and the test provider
- tests that are provided via a non-medical intermediary, such as a pharmacist or alternative health practitioner
- tests that are commissioned by the consumer but where a medical practitioner or a genetics health professional is involved in the provision of the service

The Principles address the situation where genetic tests are marketed directly to consumers rather than to qualified medical professionals.

The Principles are intended to cover all aspects of direct-to-consumer genetic testing services, including the marketing and advertising of tests, the collection, analysis and storage of biological samples, the interpretation of results and the provision of results to the consumer.

Although these Principles are intended to be applied as broadly as possible, it is also recognised that, depending on the nature of the genetic test, not all principles will be applicable in all circumstances (see 'how to use the principles' section of this document).

These Principles do not cover genetic testing carried out purely for medical research purposes, approved by a Research Ethics Committee (REC) where the results of the genetic test are not disclosed to the consumer, genetic tests for forensic purposes and court ordered genetic relatedness testing.

Table 1: Types of tests covered by these principles

Genetic tests covered by these Principles may be categorised in the following way:

1	Diagnostic tests	Tests intended to diagnose a medical condition in a person with symptoms and/or signs.
2	Pre-symptomatic tests	Tests intended to predict that an asymptomatic person has a high probability of developing a condition, for example, mutation testing in some autosomal dominant single – gene disorders. This is sometimes referred to as predictive testing.
3	Carrier testing	Tests intended to show that a person is a carrier of a condition, so that although they are not themselves affected, there is a risk they may have affected children with another carrier.
4	Pharmacogenetic tests	Tests intended to predict the response profile of an individual to a drug or course of therapy.
5	Susceptibility/Pre-	Tests intended to provide an indication of the

	dispositional health tests	absolute lifetime risk and/or relative risk of an individual developing a condition compared with the general population.
6	Lifestyle/ behavioural tests	Tests intended to provide information about an individual's: • behavioural propensities • performance capacities (physical or cognitive) or • response to certain environmental conditions and that are designed to assist the individual to modify the outcomes of any of these by elective changes in behaviour (not including the administration of prescribed medicines).
7	Nutrigenetic tests	Tests intended to provide information about how an individual metabolises nutrients.
8	Phenotype tests	Tests intended to provide information about how an individual's phenotype is conditioned by their genotype, for example, tests that indicate the genetic basis of a person's eye colour
9	Genetic relatedness tests	Tests intended to determine/ or provide information about a genetic relationship, including paternity and maternity tests.
10	Ancestry tests	Tests intended to provide information about an individual's relatedness to a certain ancestor or ancestral group and /or how much of an individual's genome is likely to have been inherited from ancestors from particular geographical areas or ethnic groups.
11	Genetic matching	Tests intended to determine the likelihood that an individual is the source of a sample of DNA recovered from a secondary object or material.

Definitions

Terms in these Principles are used with the following meanings unless otherwise indicated or determined by context:

'Genetic test' – a test to detect the presence or absence of, or a change in, a particular gene or chromosome or a gene product or other specific metabolite that is primarily indicative of a specific genetic change.

'Genetic test in the context of inherited or heritable disorders' – a test falling into any of the first three categories of table 1 (above) that is capable of providing information that may have important implications for the health of the person concerned or members of their family, or have important implications concerning reproductive choices.

‘Condition’ – a medical condition caused, solely or in part, by changes in genes or chromosomes.

‘Trait’ – any physical or behavioural feature determined, at least in part but almost always in combination with other factors, by an individual’s genotype.

‘Test provider’ – an individual, company organisation or other entity providing genetic test services to the public or a section of the public, including those services marketed or provided by an intermediary who is not a nationally recognised medical practitioner.

‘Genetics health professional’ – a clinical geneticist, genetic counsellor, medical practitioner or any other health professional who has undergone appropriate training in the interpretation of genetic information and has achieved the required competencies.

How to use the Principles

The Principles are set out in the order in which they are expected to become relevant in the testing pathway.

Insofar as they are relevant to the provision of the test to a particular consumer, the principles should be applied to the provision of ALL genetic tests supplied directly to the consumer, regardless of the category that a test falls into or the nature of the information that the test may provide. However, some tests – generally genetic tests in the context of inherited or heritable disorders – should only be provided to consumers with individualised pre- and post-test counselling (see principle 1.3). In addition, with other tests that are not genetic tests in the context of inherited or heritable disorders, but where the results are likely to have a significant or detrimental impact on the consumer, the test provider should consider whether the test results should be provided only in the context of a consultation with a suitably qualified genetics health professional, and make provisions accordingly (see principle 10.1).

The consequences for an individual of taking a genetic test depend not only on the nature of the test and the information it can reveal but also on the personal and familial circumstances of the individual taking the test. A test provider should therefore consider both the type of test being provided and the impact of that test when offering genetic tests directly to consumers and, where they are offered, in providing consumers with the results of those tests.

Principles for the provision of genetic testing services directly to the consumer

1. Purpose and scope

1.1 These Principles are intended to ensure good practice in the provision of genetic testing services directly to the consumer. The test provider should strive to provide a high-quality service that meets the expectations of the consumer whilst safeguarding their interests.

1.2 The Principles apply to tests marketed to or ordered directly by a consumer or by a non-medical intermediary acting on the consumer's behalf; they are not intended to apply to tests ordered by a medical professional on biological material taken from an individual as part of a professional investigative or diagnostic procedure in respect of that individual by that professional.

1.3 Where the test is a genetic test in the context of inherited or heritable disorders, that test should only be provided to consumers with individualised pre- and post-test counselling.

2. Marketing and advertising

2.1 Where relevant, the test provider should comply with any legislation or voluntary codes for advertising of medical tests, including genetic tests or other clinical services and they should also comply with more general guidance covering consumer advertising.

2.2 Promotional and technical claims for genetic tests should accurately describe both the characteristics and the limitations of the tests offered, and the test provider should not overstate the utility of a genetic test.

2.3 Claims made about the clinical validity of genetic tests should be supported by relevant evidence published in peer reviewed scientific literature and the test provider should give standard references to this literature.

2.4 The test provider should be aware of the risk of bias when quoting evidence and ensure that evidence is presented transparently with reference to the criteria used to include and/or exclude published literature when this is cited as evidence of the applicability or effectiveness of the test.

2.5 Information about tests which are available only in the context of a consultation with a genetics health professional or are only provided to consumers with both individualised pre- and post-test counselling should make it clear that tests are available only in that context.

3. Regulatory Information

3.1 Only clinically validated genetic variants, such as single nucleotide polymorphisms (SNPs), should be used in genetic tests. The test provider should make available the evidence of the association between a genetic marker and a disease, condition or trait for the genetic tests that they supply. These associations should be published in peer-reviewed scientific journals, they should be undertaken in line with the recommendations made in the STREGA statement*, and the provider should supply standard references for these publications.

* Strengthening the Reporting of Genetic Association Studies (STREGA)- An Extension of the STROBE Statement. PLoS Medicine February 2009, Volume 6, Issue 2, e1000022

3.2 Standard statistical methodologies accepted by the scientific community should be used to calculate the risk of the disease, condition or trait, and the evaluation of the algorithms used should be made available by the test provider for standard review and scrutiny.

4. Information for prospective consumers

4.1 The test provider should supply easily understood, accurate, appropriate and adequate information to consumers before obtaining consent for a genetic test. The following should be provided:

- general information about genetics to enable a consumer to understand the scientific basis of genetic testing, the role of genes in health and disease, and conditioning phenotypes, and the technologies applied to generate the knowledge
- a clear explanation of the relative roles of genetics, environmental factors, lifestyle choices and other factors in determining health, disease and phenotype
- specific information about genetic tests offered
- information about the presentation of results in statistical form, such as relative and absolute risk assessments, so that an individual can understand test results that are provided
- information about measures taken by the test provider and laboratories to ensure the confidentiality of personal records and security of biological samples
- information about the maximum period of storage of the biological sample and personal records, and procedures for storage, transfer and disposal of biological samples and personal records
- information about whether biological samples may be used for any secondary purposes, such as additional research purposes, and about or whether personal genetic information may be passed on to third parties and, if so under what conditions

- information about procedures for handling and resolving consumer complaints
- information about the manner in which the test results will be provided and, if applicable to the genetic test, information about the requirement and cost implications of pre- and post-test counselling or the involvement of a genetics health professional
- a statement that the results of the test might be able to reveal information about genetic relationships
- information about what will happen to consumers' biological samples, and personal and genetic data, if the company ceases trading

4.2 The test provider should provide information to consumers about the association between a genetic variant and a disease, condition or trait for each genetic test that they offer in a format that is easy to understand.

4.3 The test provider should provide information about the scope of the test, its accuracy and limitations. Information about the analytical and clinical validity* of each of the genetic markers used in the test should be made available. Other factors, such as behaviour or environmental conditions, that will play a role in determining the development of the condition or trait under investigation should be listed.

* Clinical validity includes information about (1) the relationship between the genetic marker and the condition or trait and (2) test performance, which may include the following characteristics of the genetic marker: sensitivity, specificity, positive and negative predictive values, likelihood ratios and areas under the ROC curve.

4.4 The test provider should provide information about the likely outcomes of the genetic test and the decisions that a consumer may face after taking the test. They should also identify prospectively any likely further investigations that a consumer or member of their family may wish to pursue after receiving the test results.

4.5 If a test provider intends to use a consumer's biological samples and/or associated personal or genetic data for research purposes, the consumer should be informed whether the research has been approved by a research ethics committee or other competent authority, whether the biological sample and data will be transferred to or kept in a biobank or database, and about measures to ensure the security of the sample. The consumer should be informed of any risks or potential benefits associated with participating in the research.

4.6 If a test provider intends to use the results of a genetic test to make a recommendation to a consumer to purchase a therapeutic product, such as a nutritional agent or supplement, the test provider should make available information about the link between the genetic test result and the efficacy of the indicated product. The test provider should also provide information about other lifestyle choices and behavioural modifications that are known to have a preventative or therapeutic value in relation to the trait linked to the genetic markers tested.

4.7 If a test provider intends to use the results of a genetic test to make a recommendation to a consumer to alter the dosage of a medicine or to recommend alternative medicines, the test provider should make available information about the link between the genetic test result and the metabolism of the indicated medicines.

4.8 The test provider should make it clear how and whether a consumer can receive updated test results as part of the service they supply.

4.9 Where appropriate, the test provider should inform consumers about recommendations or known actions that may help the consumer to take informed decisions about their health or welfare in the light of the test results, including informed interaction with the health care system.

4.10 Where appropriate, the test provider should supply consumers with information about genetics health professionals who are able to offer further advice or support.

4.11 An appropriately qualified professional, with recognised training and qualifications, employed by or representing the test provider, who is regulated by an appropriate professional body, should be responsible for ensuring that consumers are provided with all of the information specified in this section of the Principles.

5. Consent

5.1 In providing direct-to-consumer genetic tests, the test provider should give consideration, not only to the nature of the test and the information that it generates, but also to the personal and familial circumstances of the consumer.

5.2 A genetic test should only be carried out after the person concerned has given free and informed consent. Informed consent can only be provided when a consumer has received sufficient relevant information about the genetic test to enable them to understand the risks, benefits, limitations and implications of the genetic test.

5.3 The test provider should take reasonable steps to assure themselves that a biological specimen provided for testing was obtained from the person identified as the sample provider. They should obtain a signed statement to this effect from the person buying the test.

5.4 The test provider should require consumers to sign a statement confirming that they give their informed consent to the specific genetic tests to be undertaken on their biological material. The document should record the sample provider's age and that they have read and understood the information with which they have been provided.

5.5 The test provider should retain documentary evidence of the provision of informed consent by the consumer.

5.6 Separate informed consent should be requested by the test provider before biological samples are used for any secondary purposes, e.g. research, or before any third party is permitted access to biological samples. Consumers' biological samples and personal and genetic data should only be used for research that has been approved by a research ethics committee (REC) or other relevant competent authority.

5.7 Genetic testing of individuals who lack the capacity to consent should only be carried out if testing is in his or her best interests. As such decisions will require careful consultation with family or carers and may involve health professionals and the Courts, companies offering direct-to-consumer tests should not provide tests to adults unable to provide informed consent.

5.8 Companies offering direct-to-consumer genetic tests should be aware of the laws that exist in some countries prohibiting DNA theft, which make it illegal to obtain or test DNA without the consent of the person from whom it originated. In line with these laws a test provider should make consumers aware of the law and should not perform a test if they have reason to believe that a biological sample they have been provided with for genetic testing purposes has been taken from a third party who has not given their consent for the tests to be performed. Requests to recover DNA for genetic testing purposes from secondary objects or materials should raise suspicion and should be declined.

5.9 If a company providing direct-to-consumer genetic testing services is taken over by a third party and the new parent company wishes to perform tests on biological samples, or store biological samples and personal data, that were obtained prior to the takeover then the new parent company should obtain new specific consent from the person to whom those samples and/or data relate.

5.10 With the exception of paternity tests, genetic tests in respect of children when, according to applicable law, that child does not have capacity to consent should normally be deferred until the attainment of such capacity, unless other factors indicate that testing during childhood is clinically indicated. If postponement would be detrimental to the child's health, or the management of the child's health may be altered significantly depending on the test result, then testing should be organised by a genetics health professional who has responsibility for ensuring that any medical intervention or screening indicated will be arranged and proper arrangements made for any subsequent care.

6. Data protection

6.1 Genetic information is sensitive personal data and requires the highest level of security and confidentiality. Records containing personal data and genetic information that can be linked to an identifiable person should be subject to privacy protection and security in accordance with professional guidance and applicable laws on data protection and confidentiality.

6.2 The test provider and laboratories should not release biological samples or records containing personal data and genetic information that can be linked to an identifiable person to any third party without the prior consent of the person to whom they relate.

6.3 Companies who wish to record consumers' details on to a database that will be held by the test provider, or a laboratory or professional associated with the testing procedure, should obtain prior consent from the consumers. Consent should also be obtained prospectively for consumers to be contacted in the future by these organisations or individuals.

6.4 If a test provider ceases trading, they should dispose of personal and genetic data securely or provide for transfer of responsibilities in accordance with the terms of consent given by the consumer.

7. Sample handling

7.1 The use, storage, transfer and disposal of biological samples provided for genetic testing should be carried out in accordance with applicable legal, ethical and professional standards. The nature, purpose and maximum duration of the storage should be specified.

7.2 Biological samples should be used, stored, transferred and disposed of in conditions that ensure their preservation and security.

7.3 If a test provider ceases trading, they should dispose of consumers' biological samples securely or provide for transfer of responsibilities in accordance with the terms of consent given by the consumer.

8. Laboratory processes

8.1 The analysis of biological samples for the purpose of providing genetic testing services should be provided by competent laboratories, established by accreditation or other equivalent recognition consistent with the OECD* guidelines for quality assurance in molecular genetic testing. This includes monitoring the quality of laboratory performance through proficiency testing.

* Organisation for Economic Co-operation and Development Guidelines for Quality Assurance in Molecular Genetic Testing

8.2 Genetic tests used as part of a direct-to-consumer genetic testing service should be able to identify the genotype of interest both accurately and reliably.

8.3 Laboratories should have policies in place to apply corrective measures if their performance falls outside of parameters determined by the laboratory's quality assurance programme.

8.4 Laboratory personnel should have appropriate professional qualifications that meet recognised standards, underpinned by education and training, to assure competence in laboratory procedures involved in carrying out genetic tests.

9. Interpretation of test results

9.1 Interpretation of genetic test results should be carried out under the responsibility of an appropriately qualified professional, with recognised training and qualifications, working within the standards determined by an appropriate professional body and regulated by this professional body, employed by or working on behalf of the test provider. There should be no remuneration structure in place that would allow this individual to benefit directly from any particular interpretation of the test results or the sale of any services or products related to those results.

9.2 The qualified professional responsible for the interpretation of genetic test results should ensure that the interpretation of genetic test results is accurate and take steps to ensure that these results are comprehensible to the consumer.

9.3 Where genetic test results are provided in the form of a risk assessment, the risk assessment should be based on robustly evaluated algorithms. Standard statistical methodologies should be used to convert risks reported in scientific literature to the risk of a disease, condition or trait for an individual compared with the general population risk, as well as lifetime risks or lifetime incidences. Results should make clear the distinction between relative risks and absolute risk.

9.4 Test providers should regularly review the available evidence on which their interpretation is based.

10. Provision of results

10.1 The test provider should consider the impact of the test results for the consumer. Where the results of a test are likely to have a significant or detrimental impact on the consumer the test provider should ensure that the consumer has reasonable access to appropriate support and professional advice. In particular, the test provider should consider whether the test results should be provided only in the context of a consultation with a suitably qualified genetics health professional, and make provision accordingly. In considering what additional support is appropriate, the following factors should be taken into account:

- if used for diagnostic purposes, the severity of the condition to be diagnosed and the likely impact of the diagnosis on the person diagnosed
- if used for predictive purposes, the degree of reliability of the prediction
- if the test is intended to predict the development of a progressive disorder, the likely speed of degeneration
- the potential for the test to have a significant impact on the clinical management of the individual taking the test

- the potential of the results of the genetic test to have a significant or life-altering impact on the behaviour of the individual taking the test
- the potential for the results of the genetic test to provide health-related information about a third party, such as a biological relative
- the potential for the test to provide genetic information about a fetus
- whether the test is a stand-alone test or if there are other confirmatory tests
- the potential of the test to have a significant impact on personal relationships and the stability of families

10.2 The results of genetic tests and the significance that should be attributed to a particular genetic test result should be described to the consumer in a format that is easy to understand.

10.3 When testing for a condition or trait, where such conditions or traits are determined, at least in part, by other, non-genetic factors in addition to genetic markers, the test provider should make consumers aware of these other factors when providing results of genetic tests. In addition, the test provider should supply an indication of the level of significance that an individual should attribute to the genetic test results in comparison with the significance of these other factors, and this should be provided to the consumer in a format that is easy to understand.

10.4 The test provider should take care not to overstate the value or significance of the results of the genetic test when providing the test results.

10.5 The test provider should state clearly when a genetic test result can only give an indication of relative risk in relation to the general population as opposed to an absolute risk that might only be calculable in the context of a family history analysis.

10.6 The test provider should have in place a process to evaluate how well consumers are able to understand the background information and test results they have received, and take steps to improve their information and results provision in accordance with the findings.

10.7 The test provider should ensure that the provision of genetic test results is undertaken in such a way as to retain the confidentiality of personal and genetic data. When genetic test results are provided electronically, the test provider should ensure that appropriate security measures are in place to maintain the confidentiality of data transmitted. If the option of sending test results via email is offered by the test provider, consumers should be made aware that this method is generally not secure.

10.8 The test provider should not release genetic test results to any third parties, including insurance companies, genetics health professionals or other medical practitioners without the specific prior consent of the sample provider.

10.9 Test providers who interpret un-interpreted data obtained from genetic tests that have been provided by a third party laboratory should comply with all the aspects of these Principles that are relevant to the services they provide. Likewise, test providers who only undertake the genetic analysis and do not interpret the test results should comply with all the aspects of these Principles that are relevant to the services they provide.

11. Continuing support

11.1 The test provider should be able to provide consumers, at the time of testing or at any subsequent stage, with information about opportunities that are available for any further consultation with genetics health professionals.

12. Complaints

12.1 The test provider should have written procedures in place for acknowledging and investigating complaints. Staff who manage and respond to complaints should have received appropriate training.

12.2 The test provider should nominate a member of staff to oversee the handling of complaints. This person should be responsible for the management of the investigation of the complaint and the effective operation of the complaints procedure.

12.3 The complaints procedure and the name and contact information of the person to contact regarding complaints should be easily accessible to consumers. This information should also be available in formats suitable for people with physical or sensory impairments.

12.4 The test provider should ensure that complaints are dealt with in a reasonable time-period and consumers should be informed promptly of the outcome of the complaint.

12.5 If a consumer remains dissatisfied with the investigation or outcome of their complaint, they should be made aware of what further recourse might be available to them.

Consultation Questions

Questions in relation to the levels of support that should accompany genetic testing

1. **Do you believe that recommending individualised pre- and post-test counselling to accompany genetic tests in the context of inherited or heritable disorders is the right approach?**

   Principle 1.3 states that, “where the test is a genetic test in the context of inherited or heritable disorders, that test should only by provided to consumers with individualised pre- and post-test counselling”. Genetic tests that would evoke this requirement are tests that are capable of providing information that may have important implications for the health of the person concerned or members of their family, or have important implications concerning reproductive choices. Generally, this would be tests falling into any of the first three categories of table 1 (diagnostic, pre-symptomatic and carrier tests).

2. **Do you believe there are certain genetic tests that should not be offered direct-to-consumers? If so, which categories of tests?**

   The Principles do not suggest that certain genetic tests should not be offered direct-to-consumers. However, they do state the appropriate levels of support that should accompany testing.

Questions in relation to stratification of the principles

3. **Pre-symptomatic and susceptibility/pre-dispositional health tests are distinct categories in the draft of the Principles. Do you believe that this distinction is both valid and robust? If not, do you believe these two groups of tests could be stratified better?**

   Categorisation of genetic tests can be found in table 1 on pages 5 and 6. It should be relatively easy for test providers, regulators and consumers to determine the category of a test that is either very strongly (pre-symptomatic tests) or very weakly (susceptibility tests) able to determine whether an individual will develop a condition: such as BRCA1 and 2 tests that are able to strongly determine the development of breast cancer and the T allele of rs3803662 located on chromosome 16q12 is only weakly able to determine the development of breast cancer. However, genetic tests that have a moderate ability to predict the development of a condition such as CHEK2 mutations that are associated with a 13% lifetime risk of breast cancer may create confusion and in turn lack of consistency amongst commercial

providers as to the categorisation of tests. An approach might be to suggest a penetrance of 5% or more as an appropriate threshold for pre-symptomatic tests.

4. **Should the Principles recommend that pharmacogenetic tests only be provided to consumers with individualised pre- and post-test counselling and should they fall into the bracket of 'genetic tests in the context of inherited or heritable disorders'?**

   The Principles classify 'genetic tests in the context of inherited and heritable disorders' as a test falling into any of the first three categories of table 1 (diagnostic, pre-symptomatic and carrier tests) that is capable of providing information that may have important implications for the health of the person concerned or members of their family, or have important implications concerning reproductive choices. The Commission and working group are undecided as to whether pharmacogenetic tests should also generally fall into this bracket.

   An individual may wish to purchase a direct pharmacogenetic test as they are aware that taking a particular medicine in the past has had either no therapeutic effect or adverse side effects. A genetic test may help to establish if these effects can be explained by genetic factors. In addition, the pharmacogenetic test may not be available via the health service or consumers health insurance scheme. A risk associated with taking a pharmacogenetic test, without involvement of a medical practitioner, would be that an individual takes a test and then, on receipt of the results of the tests, decides either to self administer a drug or alter their prescribed medication. Such behaviour could have serious consequences. In some countries, the medication may only be available in the first place by prescription after a consultation with a medical practitioner; in other countries, medicines may be readily available over the internet or from pharmacies without prescription.

5. **Are the impact criteria listed in Principle 10.1 (in addition to the categorisation of tests) a helpful additional way of stratifying genetic tests? Should a list of tests be included in the Principles that determine to which genetic tests the application of principle 10.1 is relevant?**

   Test providers are expected to consider the impact of the test result for the consumer. Where the results of a test are likely to have a significant or detrimental impact on the consumer the test provider should ensure that the consumer has reasonable access to appropriate support and professional advice. In particular, the test provider should consider whether the test results should be provided only in the context of a consultation with a suitably qualified genetics health professional, and make provision accordingly. A list of factors is included under principle

10.1, which the test provider should take into account when considering the impact of the test results for the consumer.
There is potential for lack of consistency in the provision of tests as there may be differences of opinion of how test results may affect an individual. Including a list of tests that clearly identifies tests to which the application of principle 10.1 is relevant, should promote consistency. However, a list may quickly become out of date in this rapidly developing field.

6. **Are there any principles that are applicable to certain genetic tests that you consider should not be applied to that test? Specifically, do you consider the amount of information that test providers will be expected to provide to consumers to be excessive for some tests?**

Is it necessary for test providers to provide all the following information to consumers regardless of the tests they offer:

- specific information about genetic tests offered
- information about the presentation of results in statistical form, such as relative and absolute risk assessments, so that an individual can understand test results that are provided
- information about measures taken by the test provider and laboratories to ensure the confidentiality of personal records and security of biological samples
- information about the maximum period of storage of the biological sample and personal records, and procedures for storage, transfer and disposal of biological samples and personal records
- information about whether biological samples may be used for any secondary purposes, such as additional research purposes, and about or whether personal genetic information may be passed on to third parties and, if so, under what conditions
- information about procedures for handling and resolving consumer complaints

In addition, test providers will have to employ an appropriately qualified professional, with recognised training and qualifications, who is regulated by an appropriate professional body to take responsibility for ensuring that consumers are provided with all the information listed in section 4 of the Principles. Do you believe this is necessary for all genetic tests?

Questions in relation to consent

7. **Should principle 5.10 be included? (Genetic testing of children)**

   Some people take the view that this principle expresses a degree of genetic exceptionalism: as parents are able to discover many other types of information about their child and can take other decisions on behalf of their child, why deny them the same rights to find out about their child's genome? Currently there are very few adult onset conditions that are known to have a proven link to childhood behaviour; however, if such links were discovered in the future, genetic testing of a child would give a parent the chance to modify the management and upbringing of a child to take into account genetic predispositions that may have the effect of discouraging the onset of disease in later life.

   Others take the view that the autonomy of the future adult should be protected, so that the future adult can make their own decisions about taking a genetic test when he or she reaches an age when he or she can fully understand the implications for genetic tests and decide in accordance with their own values (the exception to this is the case when not taking a genetic test could be detrimental to the health of the child). There are also concerns that having genetic information about a child could result in a parent putting additional pressures on a child to pursue certain directions to the exclusion of others.

8. **Principle 5.3 states: "The test provider should take reasonable steps to assure themselves that a biological specimen provided for testing was obtained from the person identified as the sample provider. They should obtain a signed statement to this effect from the person buying the test"**
   What do you consider to be 'reasonable steps' and should the Principles state what these steps should be?

   It is difficult to ensure beyond doubt that a biological sample is from a certain individual. However, we do believe there are some measures that test providers could adopt to improve their knowledge of the individual they are testing and to discourage DNA theft. Such measures would also highlight the importance of consent to the consumer, who may have been previously unaware of this. For example, the test provider might request consumers to have the taking of biological samples witnessed by a 'person of recognised standing' in order to confirm the identity of the individual. This might make testing more costly and less convenient for the consumer and may not fulfil its purpose very effectively in cases of deliberate identity fraud. However, it would raise the importance of this issue to both test providers and consumers.

9. **After discussions within the working group the following principle was not included: "A test provider must take whatever measures**

are necessary and appropriate to ensure that an individual has provided informed consent and has capacity to provide that consent for a genetic test." Do you think this principle should or should not be included?

Such a principle would expect test providers to have a dialogue with the consumer, prior to testing, to establish that the individual has understood the information they have received or read about the test. As the Principles recommend that certain genetic test results should only be provided either with individualised pre- and post-test counselling or within the context of a consultation with a genetics health professional, the principle above may not be considered necessary. For tests where the results have the greatest potential risks, a genetics health professional will be able to explain the test results to the consumer and ensure they understand the significance of any results. If the requirements for pre- and post-test counselling and the involvement of a genetics health professional (principles 1.3 and 10.1) are removed, then there may be a greater need for this principle.

Other questions

10. **Are any of the principles impossible to apply in your jurisdiction given existing national legislation or regulatory constraints?**

    The Principles have been written broadly so that they can be applied across different jurisdictions. If we have overlooked legislation or regulation that exists in your jurisdiction, we would appreciate details of this legislation or regulation or website/contact details where we can obtain this information for ourselves.

11. **Do you believe that test providers should sign up to the Principles and what costs do you expect will be incurred by complying with the Principles?**

How to respond to the consultation

Responses are welcomed from any individual, group or organisation from any country.

Form of responses

Responses may be brief or lengthy and may comprise comments on any areas of the Principles, including the structure of an individual principle, whether a principle will fulfil its purpose or if in fact a principle should actually be included, as well as comments on any other significant issues that have not been covered by the Principles that you feel should be included. In particular we would welcome responses to the consultation questions we have set out as these are areas that have provoked the greatest levels of debate both within the working group and the Commission. Responses need not address all the questions that are set out in this document and an individual/group/organisation may not feel that they have sufficient knowledge to respond to all of the questions.

As with all consultations, the most helpful responses will be those that set out the reasons, arguments and evidence that have led to or support your conclusions.

How to submit a response

Responses may be submitted in any form but, for convenience and ease of reproduction, we especially welcome responses by email. Response should be sent

by email to: PrinciplesConsultation@dh.gsi.gov.uk

by post to:
Principles Consultation
Human Genetics Commission
Department of Health
6th Floor North
Wellington House
133-155 Waterloo Road
London SE1 8UG

Information about respondents and publication of responses

It is helpful – although not essential – for us to know your name and contact details (email or postal address), and whether you are responding on your

own behalf as an individual or on behalf of an organisation. It is also helpful to us if you can indicate the nature of your interest in this consultation.

You should also indicate whether you are content for your response to be made public. The content of all responses will be made openly available following the consultation unless the respondent has specifically requested that it be kept confidential. Our usual practice is to publish the names of the organisations that have responded but not the names of individual respondents or any elements of responses that contain personally identifying information. All responses may be subject to requests made under the UK Freedom of Information Act 2000.

Receiving a copy of the final version of the Principles

If you wish to receive notification when the final version of the Common Framework of Principles for direct-to-consumer genetic testing services is published, please include your name and email or postal contact details.

Consultation period

The consultation period will run from 8 September 2009 to 6 December 2009 (a period of 3 months). All responses received by the HGC before the 6 December 2009 will be taken into account by the HGC's Principles working group in developing a final version of the Principles.

Further information

Further information is available from the HGC and up-to-date information about the progress of the consultation will be posted on the HGC website.

website: http://www.hgc.gov.uk/Client/Content.asp?ContentId=816

by telephone: 020 7972 4148

by email to: PrinciplesConsultation@dh.gsi.gov.uk

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