

Response to HGC Consultation Paper

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

This is a response from the Economic and Social Research Council (ESRC) Centre for Social and Economic Research on Innovation in Genomics (ESRC Innogen Centre), based at the University of Edinburgh and the Open University. The centre, which began in 2002, focuses on research that connects the social with the life sciences. The life sciences have the potential to transform health care and food production systems in developed and developing countries and to provide one of the main platforms of economic growth and global competitiveness in the 21st century. Rapid developments in life sciences challenge our existing regulatory systems and raise new ethical and social issues. Innogen's research aims to provide a sound base for decision-making in science, industry, policy and public arenas related to innovation in life sciences. Staff at the Innogen Centre have, for over twenty years, researched issues of regulation, innovation and engagement in the life sciences (both agricultural and health related), but also in other sciences and technologies of information, communication, energy and environment. The researchers working at Innogen include social scientists, economists, and lawyers. Innogen also engages with a wide range of stakeholders, nationally and internationally, including scientists, industry and private interest groups, policy makers and regulators, and citizens and public interest groups.

Summary of response

The following answers/comments to some of the HGC's principles and consultation questions draw on Innogen's research and expertise in social, economic and legal issues around genomics and public health. We have answered only those questions where we have specific data/expertise to make an informed and meaningful contribution to the consultation.

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Consultation Questions

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

The requirement for counselling in the case of genetic tests for heritable disorders is sound, and simply extends standard practices in face-to-face clinical contexts to the

broader area of direct-to-public genetic services. A report of the Advisory Committee on Genetic Testing (ACGT), published back in 1998, recognised the importance of providing sufficient information and counselling to potential users of genetic testing services provided outwith a formal clinical context. Although genetic counselling has been subject to much sociological critique in the past for its failure to be non-directive and its potential to actually undermine individual autonomy and choice (see Mitra, 2007), it would seem that some form of knowledge dissemination and counselling ought to be a prerequisite for the marketing of genetic tests direct to the public; particularly in the case of heritable diseases with strong penetrance and health implications for other family members. Nevertheless, counselling should not be considered a panacea for the many challenges facing direct-to-consumer genetic testing.

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

In principle, there does not appear to be sufficient justification for restricting the range of genetic tests offered to consumers, so long as the test is clinically validated, based on the best available scientific evidence; and the information and counselling services provided are appropriate to the specific test being marketed. A rigorous process for validating different types of genetic test would seem vital.

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?

It would seem appropriate to differentiate tests based upon their “predictive value”, as this Principle seeks to do. The regulatory framework and level of information/counselling provided to the user may need to be tailored to *both* the predictive efficacy of the test and the severity of the condition being tested for. However, it is important to recognise the limitations of gene penetrance studies. In 1997, a report by the Human Genetics Advisory Committee concluded:

“It’s one of the basic misunderstandings of genetics that it will lead to an immediate increase in general predictive power. These expectations seem unrealistically high ... it is unlikely that actuarially important genetic predictions of common causes of adult death will be available and validated for some time to come. This is because the information linking genetics and multifactorial disease is at too early a stage to make sound assessments of added risk.”
(HGAC, 1997, paragraph 2.12).

This statement appears to support the HGC’s Principle that a distinction should be made between tests with “strong” and “weak” associations. However, even tests for genes with supposedly strong penetrance have been questioned. The empirical problem of predictive efficacy was dealt with comprehensively in a 2001 Parliamentary Science and Technology Committee Report (HOC, 2001) investigating the relevance of genetic tests for insurance. During the oral evidence sessions, Prof Marin Bobrow (Head of clinical genetics at Cambridge University) challenged the orthodox belief that the genetic test for Huntington’s Disease (a rather simple, single-gene disorder) had a high predictive value. In particular, he stated that the age-of-onset could vary considerably. The Committee report concluded that genetic tests (even those for supposedly simple, single gene disorders) could indicate presence or absence of a gene defect, but could not predict with any certainty whether or when it

would be expressed. Therefore, we must be careful not to reify genetic exceptionalism and uncritically bestow genetic tests with undue predicative efficacy (see Mitra, 2006). In relation to the specific question, a distinction should be made between genetic tests, but the predictive value of *all* tests must be constantly evaluated in light of best available scientific and clinical evidence.

4. Should the Principles recommend that pharmacogenetics 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'?

Due to the potentially serious consequences of a patient misusing the information derived from a pharmacogenetic test, as illustrated in the consultation document, it would be prudent to treat such a test on the same basis as a heritable genetic test. Individual pre- and post-test counselling should be a key requirement. Ideally, pharmacogenetic tests should only be administered in a clinical context, but if they were to be provided directly to the public, it is vital that patients are given the requisite background knowledge and counselling to use the information in an appropriate manner.

6. Are there any principles that are applicable to certain genetic tests that you consider should not be applied 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?

Although the range of genetic tests are quite diverse in terms of their predictive efficacy, severity of penetrance and implications for personal health management; it would not seem prudent to apply different standards for the provision of information and data handling. First, consumers should be informed about the nature of the test, its clinical relevance and validity; and statistical interpretation regardless of the nature of the genetic test. Furthermore, it is clear from our research, and that of other social scientists, that patients and publics expect to have some control over the use of their personal biological information and tissue samples. Issues of informed consent and privacy over genetic data are complex and need to be handled sensitively if the public is to feel confidence in genetic testing procedures. People are concerned about potential commercialisation of their personal data and misuse of tissue samples (see Haddow, 2007 and Laurie, 2002). Therefore, providers of all genetic testing services to the public should explain exactly how the information will be stored and handled. In the absence of this requirement, there would be a real risk of serious misuse of data.

In relation to the question about test providers employing an appropriately qualified professional to ensure the information is disseminated correctly, this would indeed be sensible. It would also be necessary for such a professional to apply the same standards to all genetic tests, for the reasons outlined above.

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?

In light of our previous comment that informed consent and trust in the storage, handling and use of genetic material is crucial for the safe provision of genetic testing

services direct to the public; all reasonable steps should be taken to ensure biological samples originate from the individual seeking to be tested. We are not qualified to advise on what exactly the reasonable steps should be, but accept the argument that if the requirements are too onerous and expensive, it could have negative commercial implications. There needs to be an appropriate balance so that the commercial provision of testing services is profitable whilst at the same time ensuring the highest possible standards of privacy are met. The Principles should, in our opinion, attempt to set out what some of the key steps should be, and at least go some way to highlight the importance of the issue for both providers and consumers of genetic testing services.

9. After discussions within the working group the following principle was not included: “A test provider must take whatever measures are necessary 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?

Notwithstanding the fact that the level of informed consent will be partly dependent on the individual consumer, and it is always difficult to accurately define “informed” in this context, providers of genetic testing services should take all reasonable measures to ensure individuals have been given sufficient information to make an informed decision. This may include simply signing a consent form or talking face-to-face with a healthcare professional. For those tests that require pre- and post-test counselling, this is not a problem, as the consultation document recognises. However, just because some genetic tests will require genetic counselling does not mean that this principle is then redundant. Regardless of the test being offered, there needs to be some form of “informed consent”, and the provider ought to take all reasonable steps to achieve this. Therefore we would suggest that this principle be reinstated. If nothing else, it would serve to highlight the importance of informed consent and place an onus on the provider to ensure consumers are armed with as much information as necessary prior to undergoing a genetic test.

References

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