

Public consultation: Human Genetics Commission

Response prepared December 2009 by Dr Christine Knight (Policy Research Fellow) and Dr Steve Sturdy (Deputy Director) on behalf of the ESRC Genomics Policy and Research Forum.

The Genomics Policy and Research Forum is a novel ESRC-funded initiative dedicated to the development of links between social scientists and scientists working in the contemporary life sciences, and the connection of research in this area to policymakers, business, the media and civil society. The Genomics Forum is based at the University of Edinburgh and is part of the ESRC Genomics Network (EGN), a major ESRC investment spanning five of the UK's leading universities examining the development and use of the science and technologies of genomics.

Question 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?

Individualised pre- and post-test counselling to accompany genetic tests in the context of inherited or heritable disorders is a prerequisite for the ethical and effective delivery of such tests. For this reason, such tests are best delivered through face-to-face personal healthcare services, and by recognised and qualified healthcare professionals. Where direct-to-consumer genetic tests are concerned, the providers of such tests should be urged or required to provide such counselling themselves, or to refer their users to appropriate providers. However, further thought will need to be given to determining just what form such counselling should take, and how the recommendation to provide it should be policed and, where possible, enforced. This is particularly relevant where direct-to-consumer tests are provided via the internet, where consumer and test provider may be geographically distant and face-to-face counselling therefore difficult to deliver.

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

By and large, such genetic tests as have been clinically validated and are seen to provide information of significant benefit to users are available through the mainstream healthcare services, along with appropriate counselling. There is therefore little medical or commercial reason why such tests should also be provided direct to consumers, and arguably good reasons why they should be banned, especially where counselling is not provided at the same time. Nonetheless, such tests are often included in commercial packages of the kind provided by 23andMe. Given the way that such packages are marketed and delivered over the internet, it is not clear that an outright ban could be enforced; more effective might be to urge the voluntary withdrawal of such tests as are already available through mainstream healthcare services.

In contrast, most tests on offer from direct-to-consumer providers are precisely those that are not available through the usual healthcare channels. These can in turn be divided into two further classes. First there are tests for inherited disease predispositions that either have not been widely validated, or where only limited preventive or therapeutic interventions are available for the conditions indicated. While the clinical value of such tests is nugatory, there may nevertheless be significant consumer demand for their provision. As with other medical tests, it is difficult to see that an outright ban would be either necessary or enforceable. Rather, providers should be urged or required to provide clear information about the limitations of such tests, and where appropriate to provide counselling or to refer their users to appropriate providers, as already discussed in relation to Question 1 above.

Finally, direct-to-consumer genetic tests include a variety of tests for ancestry and for non-disease conditions such as drug response and eye colour, which on the whole do not have implications for health. There is little reason to prohibit the provision of such tests. Here too, however, consumers should be given clear information and guidance regarding the limitations of such tests. This would be valuable in the case of ancestry tests, for instance, when the personal identity issues invested in such tests may far exceed the meaning of the tests themselves.

Question 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?

In our view the distinction between pre-symptomatic and susceptibility/pre-dispositional health tests is problematic. The notion of a 'pre-symptomatic' genetic test implies that a condition is already present but asymptomatic or latent. While tests for gene expression may in time make it possible to diagnose such conditions, simple genetic test cannot do so; at best, some tests can indicate with 100% certainty that certain conditions will develop within an individual's anticipated lifetime. Even then, there may be considerable variation in the time of onset and severity of the condition. Susceptibility or pre-disposition, on the other hand, could in principle mean *any* degree of likelihood that an individual may develop a particular condition in the future. Insofar as the distinction between pre-symptomatic and susceptibility/pre-dispositional genetic tests might have some value, the designation 'pre-symptomatic' should therefore be reserved for single-gene conditions where someone with the relevant gene will definitely develop that condition at some subsequent time. Clarity and accuracy in using the term 'pre-symptomatic' may be particularly important in light of concerns that consumer anxiety and fatalism may result from genetic test results and the way these are reported.

Question 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 Commission's concerns around direct-to-consumer pharmacogenetic tests relate principally to the risk 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', with possible serious health consequences. This risk is a real one. However it is not clear that it would be best addressed through individualised genetic pre- and post-test counselling such as is relevant to inherited or heritable disorders, except where the tests in question *also* (coincidentally) provide information about inherited or heritable disorders. In other cases, what is required is appropriate follow-up care and advice by a medical professional qualified to prescribe the drug in question, in order to discuss with the patient the implications of self-administration or alteration of a drug regime, and to provide appropriate medical opinion and advice. As with genetic tests for inherited or heritable disorders as discussed under Question 1 above, the providers of direct-to-consumer pharmacogenetic tests should be urged or required to provide appropriate follow-up care themselves, or to refer their users to appropriate providers.

Question 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?

In our view, the impact criteria listed in Principle 10.1 provide a very useful set of criteria for considering the circumstances under which different tests should be delivered. It is not clear that this would provide a means of "stratifying" tests, so much as a set of guidelines for evaluating tests on an individual basis. Nor is it clear that the Principles should include "a list of tests ... to which ... the application of principle 10.1 is relevant". Principle 10.1 is relevant to the evaluation and application of *all* genetic tests. Rather, what might usefully be included in or appended to the Principles is a list of tests which, when evaluated against the criteria listed in Principle 10.1, should only be provided under certain circumstances – for instance with guidance from a genetic counsellor or genetic pharmacologist, or with additional information

about the unvalidated status or other limitations of such tests. Any such list should not be exclusive, and this should be clearly indicated in the Principles. In addition, the consultation document rightly recognises that such a list may quickly become out of date in this rapidly developing field. In order to address this it would be important that such a list be maintained as an annex to the Principles, or indeed as a separate register, which is regularly updated, along the lines of the Human Fertilisation and Embryology Authority register of conditions licensed for pre-implantation genetic diagnosis.

Question 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?

Given the uncertainty surrounding the significance and interpretation of many of the direct-to-consumer genetic tests currently available, and concerns about the uses to which consumers' genetic data may subsequently be put by the company providing those tests, it is vital that consumers be provided with adequate information both to make sense of their results and to make an informed decision whether or not to undergo testing. The kind of information listed at section 4 of the consultation document provides a useful guide in this respect. However, further thought should be given to exactly how that information should be framed and presented. There is a risk that the amount of information required to be provided will be overwhelming for consumers, and may even be counterproductive, in that consumers will be unable or unlikely to properly assess the information and make an informed choice about testing. There is also a risk that such information will unwittingly confer an appearance of medical significance on tests that might be better regarded as purely recreational; so far as possible, the amount of information provided should be no more than is warranted by the importance or significance of the tests themselves.

We note also that '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'. We have no difficulty with this requirement in principle; however it is not clear on the face of this statement what kind of 'professional' is required. For example, this could refer to a qualified geneticist, other medical professional, or indeed a communications or public relations professional. If this requirement is carried forward then this must be clarified in the Framework.

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

In our view the more important principle to be balanced here is the autonomy of the future adult, so that principle 5.10 should indeed be included. In addition, the principles should clearly spell out that no genetic testing of children should be carried out by direct-to-consumer providers. If a genetic test for a child is indicated for health reasons (and not taking a genetic test could be detrimental to the health of the child), then a direct-to-consumer provider must refer the family to an appropriate medical provider within a regulated healthcare framework.

Question 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?

As the consultation document notes, it is difficult to ensure beyond doubt that a biological sample is from a certain individual. This is particularly difficult in the context of online provision of genetic tests, where consumer and provider may never meet in person. Requiring consumers to have the taking of biological samples witnessed by a 'person of recognised standing' (eg, Justice of the Peace) in order to confirm the identity of the individual may indeed be ineffective in cases of deliberate identity fraud, again especially given the likely geographical separation of consumer and provider in cases of online testing provision. However, this would be parallel to requirements for proving identity in other contexts (for instance, provision of a statutory declaration in relation to pension or other financial decisions), and short of

outlawing online genetic test provision entirely (which might only encourage a black market), would therefore seem to be the appropriate legal mechanism, albeit an imperfect one.

Question 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?

Again, in an internet context where consumer and provider never meet face to face, it is difficult to imagine what measures might be taken to ensure informed consent and capacity to provide that consent. However, as indicated in the consultation document, 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. Nonetheless, we remain concerned that there is insufficient research and understanding at this point of how, and why, consumers are using direct genetic testing services. In the absence of an understanding of (for example) whether consumers mainly use these tests for recreational, genealogical or medical purposes, it is very difficult to know how stringent requirements for ensuring informed consent need to be.

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

Question 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?

As noted at Question 6 above, the amount of information that test providers will be expected to provide to consumers does certainly seem to be extensive. The burden on providers must be considered, and in principle should be roughly equivalent to the burden on medical providers advising patients within a regulated healthcare context.

Other comments

As noted above at Question 9, we remain concerned that there is insufficient research and understanding at this point of how, and why, consumers are using direct genetic testing services. This has implications not only for many of the suggested Principles, but also for whether or not to regulate (or provide a quasi-regulatory code of principles) in this area at all. The authors of the present response do not hold a settled opinion on this latter point. However, we recommend that the Commission should give due consideration to the possibility that regulation might be best delayed until we have a better knowledge of consumer behaviour in this area, and the effects of direct genetic tests on consumers.

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