

This response was submitted to the consultation held by the Nuffield Council on Bioethics on *Medical profiling and online medicine: the ethics of 'personalised' medicine in a consumer age* between April 2009 and July 2009. The views expressed are solely those of the respondent(s) and not those of the Council.

## **Response to Nuffield Council on Bioethics consultation**

### **Question 2 Validity of information**

*While much health related information is freely available to individuals, this varies greatly in quality and accuracy. Many of the lifestyle and health books and magazines that are currently available may contain medical information that is misleading or even incorrect from a scientific point of view. Do you think that information provided by DNA profiling and body imaging services raises different questions and should be subject to different regulations?*

#### **Response**

My response arises from five years of research on the regulation of pharmacogenetic tests and genetic tests for common complex diseases. I attach several publications which outline the key issues and proposals which have been developed from this research programme in collaboration with colleague's including Gail Javitt, David Melzer, Kathy Liddell, Tom Ling, Paula Saukko and Ron Zimmern

DNA profiling services may be subject to two regulatory regimes – for laboratory quality assurance and for medical devices. Both regimes may provide a measure of protection for the public from the possibility of erroneous results or misleading claims.

Regarding restricting access to services, there are two levels of restriction. One might restrict access by requiring that a test can only be ordered by a healthcare professional, in effect making it a 'prescription only' test or one might ban a test altogether. Regarding the former option, as with pharmaceuticals it is possible to envisage a mixed economy where some tests are available direct-to-consumer and others are not. This seems a more balanced approach than simply suggesting that no tests be available DTC or that any restrictions should be rejected on the grounds that they would be paternalistic. What might be restricted as 'prescription only' is an issue which is addressed in the forthcoming 'Guiding Principles' being developed by the Human Genetics Commission working party (see further discussion below).

Regarding tests which "have limitations in terms of scientific validity and clinical value" the main thing which may need regulating is the promotional claims which the companies make. Such claims can be regulated under the regulatory regime for IVD devices – in Europe the IVD Directive (for problems with this Directive, see attached documents). It is a mistake to think that these tests should be regulated chiefly under consumer legislation. Firstly, the Directive is the key regulatory instrument for IVD tests and therefore has precedence over consumer legislation for these products, and secondly, consumer protection mechanisms are largely reactive and therefore can at best deal with problems after they have occurred, they cannot prevent them occurring.

Individual consumers may be able to exercise a range of private law rights against a product manufacturer or supplier. However, the important overview point to note is that private rights can only be exercised *after* sale/supply and after damage has occurred, and, in the first instance at least, by *individual* consumers. The compulsive force of such rights in affecting the behaviour of manufacturers and suppliers is, therefore, of somewhat limited value. Individual consumers may not be able to take adequate steps to

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protect themselves, whether because of problems over obtaining access to the legal system or because of individual incapacity or inertia. It is true that some mechanisms exist under which some collective action may be taken by a group of consumers, but these mechanisms are not well developed or effective in the circumstances under consideration. The major problem with mechanisms of this type is that they are of limited value since they always operate *post facto*. Thus, controls that operate *before* sale or supply are far preferable as tools in modifying behaviour and safeguarding consumers/users. (from a private briefing provided by Christopher Hodges, University of Oxford)

Given these limitations the use of the regulatory regimes for IVD devices is a preferable instrument. Premarket review of tests to ensure truth-in-labelling and truth-in-promotion can be carried out under the IVD Directive. This is more likely to prevent problems occurring.

However, some tests may be of such poor quality that the second form of restriction (an outright ban) may be more appropriate. Much of the current policy discussion assumes that companies are probably testing the right markers, or at least those SNPs which current scientific consensus suggests are associated with a given disease or other phenotype. However, I have heard scientists criticize some companies for offering tests where the SNPs used are not supported by the scientific literature. There is a clear need to prevent such tests being sold on the basis that they are completely worthless, misleading and not fit for purpose. Moreover, such tests illustrate the fact that banning DTC delivery and requiring the involvement of a health professional, may be of no value if the test itself is worthless. Both the companies I have heard criticized for offering such tests offer their tests through physicians. Clearly, in some cases it is not DTC provision that is the problem.

Some have suggested that it is too early to act, that this is a nascent industry, that we do not know enough about the effects of these tests on consumers and that we must not confuse traditional clinical genetics with the new kinds of services now emerging. I would agree that transposing wholesale the governance frameworks from the world of clinical genetic testing is inappropriate to testing for susceptibility to common diseases. In the world of clinical genetics, governance issues have been driven primarily by the monogenic disease model of high-penetrance genes. To discover that one has the gene for Huntington's Disease is to know that one will die of that condition if one lives long enough. A couple who discover that they are both carriers of CF genes face profound questions relating to reproductive decision-making. The familial implications of monogenic disease require careful management of the testing process.

Susceptibility testing for common conditions such as heart disease, where genetic factors reveal at most moderate risk elevation, bears little resemblance to the traditional world of clinical genetics. In most respects such tests are more akin to traditional risk predictors such as cholesterol. Hence, it is appropriate that we consider carefully whether the governance arrangements we have developed for clinical genetics are appropriate to these new tests. Do we, for instance, require detailed pre- and post-test counselling for a susceptibility test? Should the provision of such testing be restricted to a small number of laboratories, as is the case in some countries, such as Belgium?

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In fact discussion of these issues is well-established, and the need for a nuanced approach to governance is reflected in international standards such as the OECD guidelines on quality assurance for molecular genetic testing. For instance, these guidelines emphasise the importance of counselling but acknowledge that the level of support required will depend on the nature of the test. At a recent meeting convened by the Human Genetics Commission to discuss development of a code of practice for consumer genetics, there was agreement amongst industry and the other stakeholders present that a pragmatic and nuanced approach is required and that such an approach required two things: a set of minimum common requirements which all tests and testing services would be required to meet (such as laboratory quality assurance systems and the provision of accurate and comprehensive information about tests to end users) and a means to identify those tests for which stricter regulation was required (there was no support for the idea that a test for Huntington's Disease should be made available direct to consumer).

Thus the development of policy is, at least in some countries, already informed by an appreciation of the need for what we might term a risk-based approach to the regulation of genetic tests. It is time to take sensible and measured action. To fail to do so would be to indulge in a form of unnecessary genetic exceptionalism; after all if susceptibility tests are akin to cholesterol tests, then why should they not be subject to the same regulatory constraints? To require companies providing such tests to adhere to the laws and regulations governing clinical laboratories and in vitro diagnostic medical devices is not unreasonable and would provide some minimal level of protection for consumers. That such protection is not already in place cannot be explained by lack of prior discussion, because the debate about the need for enhanced oversight of genetic tests has been going on for well over a decade. Our first policy challenge is moving beyond recommendations to action to ensure this base level set of protections for consumers.

The second policy challenge will be in deciding where such minimal requirements are not sufficient and what additional regulation is required. At what point, does the degree of heritable risk identified or the potential clinical outcome, become sufficient to warrant that the test be delivered by specialists? Take for instance a genetic test for autism, which a couple might use for reproductive decision-making, or a test for breast cancer, which, if sufficiently high risks were uncovered, might lead to consideration of prophylactic mastectomy. What about a test (such as has been commercialised by Celera) which uncovers not only a heightened risk of heart disease but also information about the individual's pharmacogenetic response to statins?

Here the policy challenge is that whilst we have mechanisms for deciding which drugs should be available over-the-counter and which require a doctor's prescription; we have no equivalent for diagnostic tests. The arguments we are having are not new, controversy has flared around DTC pregnancy tests and HIV tests in past decades: what is new is the scale and pace of change. The problem is particularly acute in this field for a variety of reasons, the chief being that it is very simple to go from a new research discovery straight to commercialisation. In general the SNPs are already on the chips. Couple this technological advantage with the new medium of internet delivery as a means to engage directly with the public then you have the perfect storm for consumer diagnostics.

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It is in this regard, above all, that we are witnessing something truly novel in the diagnostics sector. The IVD industry has long had a consumer market, and a growing range of tests are available for purchase over the internet or in pharmacies, but never before have we had a situation where new biomarkers move from discovery to DTC provision with no intervening period of gradual adoption by the medical profession.

Finally, I would like to take issue with the idea that requiring a medical consultation for a genetic test infringes an individual's 'right' to have access to their genetic data – it does not: it simply defines who the gatekeepers are. DTC genetics companies are gatekeepers too; they control access by setting standards on:

- what they report
- how they report it
- who they report it to
- how much it costs (price is a gatekeeping mechanism) and
- by keeping competitors out of the market.

The debate is not about whether we have unfettered access to our genomes - only a handful of scientists with the correct training and access to the necessary equipment might be said to have such a privilege – it is about who are the gatekeepers and what sort of controls are in place.

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