

Response to consultation: *Supply of genetic tests direct to the public*

Introduction

McFarlane Valentine BioConsulting (MVBio)¹ specialises in advising legislators, strategists and administrators formulating public policy and industrial strategy in the fields of biomedicine and agricultural biotechnology.

In brief, we support different levels of regulation for different levels of predictability. Consequently we advocate option 3 (voluntary regulation with restrictions on the type of tests that can be offered: some tests requiring access through a registered health care provider, others available subject to a voluntary code of practice) as set out on page 23 of the consultation document². This, and other opinions below, are our personal views and should not be taken to represent the position of any of our clients or partners.

What does a genetic test offered direct to the public mean?

We agree with the definition of genetic test in the first sentence of para 29, but with the proviso that this includes protein-based tests too. In particular, we believe that the desire for robust tests will lead to a search for protein-based diagnostics (as are already used for HIV, fertility, pregnancy and PSA tests), rather than DNA-based tests. Protein-based tests can be developed in situations where specific protein reagents can be generated that discriminate among different variant forms of a protein (gene product) that correspond to different forms of the gene. Protein-based tests are commonly more easily packaged, require less interpretation and have longer shelf lives.

Sale of test kits direct to the public (Q7)

There appears to be an assumption within the consultation that testing will be provided as a service by a contractor, but perhaps the notion of test kits themselves being available to consumers (as they are for home pregnancy and fertility testing) should receive some attention. The Medical Devices Agency would presumably have jurisdiction over the accuracy and reproducibility of such kits but we are concerned that tests may still not be suitable for the stated purpose. Should a molecular biologist be allowed to order a CF test (for example) and test herself for carrier status? There will be issues of interpretation with more complex tests and we feel direct access to tests should be discouraged. However if robust protein-based tests (as in the examples above) or some form of hardy DNA chip become available this will become a very live issue. Our view is that kits that divulge predictive information about major disease should not be directly available to the public. However a do-it-yourself kit to find out if taking non-prescription tablets containing codeine is worth the money could be made available.

Why might someone want a 'direct to public' test? Is provision through private health care an alternative?

There are a number of potential reasons why a direct test might be sought including:

- Where a patient doesn't want to let their GP know because they distrust security/ confidentiality of their medical record (see section on GP record below)
- Where a particular test is not available through the NHS.

In these two cases, and rather than 'direct to consumer', we suggest that tests could alternatively be provided through a private healthcare provider employing health professionals who hold a UK professional registration (for example a medical doctor or perhaps a nurse consultant). Such practitioners would be subject to the same professional mores and regulation as NHS colleagues. If a test is offered to the public through a private healthcare practitioner then this should not be regarded

¹ See <http://www.mvbio.co.uk>

² See <http://www.hgc.gov.uk/testingconsultation>

as 'direct to the public'. This would accord with the sentiment of the previous code of practice and guidance³ (see page 7): "*this code of practice is not intended to cover genetic testing provided in the context of professional medical practice*".

The National Institute for Clinical Excellence and the Health Technology Board for Scotland should give consideration through their health technology assessment procedures as to which tests it is appropriate for the NHS to fund, and therefore to which tests private access might be needed. Recognition must however be made of the fact that much of the cost of testing lies in the follow-up rather than the initial test. Consequently a decision to not provide the test through the NHS will not save the counselling and follow-up costs if, after the test is taken outside the NHS, the patient then consults a GP or specialist NHS provider for clarification or discussion of subsequent treatment options.

- Someone seeking testing of a sample doesn't want a healthcare worker involved because the source of sample is dubious.

We imagine it might be easier to get an illicitly obtained sample derived from a colleague, employee or family member analysed if you don't have to explain yourself to a suspicious healthcare worker. This should be dealt with through the proposed criminal offence of obtaining genetic material by deception/without consent.

We support different levels of regulation for different levels of predictability (Q7)

As in many discussions of the 'awesome power of genetics' it is important to realise that sets of genes interact with each other, and with multiple environmental and social factors, to influence general health. Consequently, the information obtained from individual genetic tests, although powerful in predicting whether an individual will suffer from a condition where there is a pre-existing family history, is considerably less useful in assessing susceptibility to common causes of morbidity (disease) and mortality. These considerations mean that the need for family history is of great importance. In fact without family history it will very often not be possible to determine which genetic test is required, even where the condition in question is known. This is acknowledged at point a) on page 4 of the introduction to the current code of practice: "*Many test results are complex and cannot be interpreted meaningfully without a medical or family history, which cannot be incorporated adequately into mail-order or over-the-counter test procedures.*"

For example with the breast cancer susceptibility genes Brca1 and Brca 2 if an individual is only tested for the 'common mutations' the test may miss the inherited mutation present in their family. Testing the entire gene by complete sequencing would be possible but very expensive. Consequently, the current NHSScotland guidance in this area is that family history, and (after obtaining consent) genealogy using cancer registrations of relatives, is advised before commissioning molecular genetic tests⁴.

We would therefore argue **that where targetted genetic tests are contemplated to determine risk of serious disease or disorder these should be offered only through healthcare practitioners holding a UK professional registration**. These need not be NHS providers: current expert opinion in the US advocates private provision, but again only through registered healthcare providers.

However, there are increasing numbers of applications seeking to persuade consumers that they should modify their behaviour on the basis of a genetic predisposition. This may be valid in relation to medications, for example certain over-the-counter painkillers, where some individuals will derive much less benefit from taking a codeine containing tablet, but is less valid in the area of dietary advice since the ingested ingredients represent a much more mixed sample. In this last case, the issue is more of consumer protection: i.e. is the advertising legal, decent, honest and truthful, the limitations of the approach fully disclosed and the testee fully aware of those limitations. If after such information is fully

³ See <http://www.doh.gov.uk/genetics/hgts.htm>

⁴ See http://www.show.scot.nhs.uk/sehd/mels/hdl2001_24.htm

disclosed the testee freely enters into a contract and gives consent then we feel there need be no further specific protection.

We do not advocate statutory regulation of 'genetic tests to suggest lifestyle changes' but rather an agreed code of practice that test vendors can adhere to and use as a badge of quality and compliance.

Additionally, where a genetic test is offered to determine suitability to an individual of a non-prescription medication we believe that test should also be available 'direct to consumer', subject to compliance with a voluntary code of practice and perhaps under the supervision of a registered pharmacist.

What information and assurances might feature in a code of conduct

We envisage that testees would require information on the limitations of the test they are about to take (e.g. its predictability) and assurances about how long the sample they provide would be kept and who might access the information derived from the test result.

Requirement for throughput?

For testing as part of national screening programmes there is currently a requirement that laboratories carry out a minimal throughput of a particular test in order to ensure continued familiarity. There might require to be mention of this in information provided to prospective testees in order to ensure confidence in competence.

Who decides which tests require regulation?

Our neat division does not cater for what is in practice more of a spectrum - who would decide which tests have heavy consequences? Perhaps determination of which tests lie within the more restrictive category could be through an expert advisory committee (modelled on the Genetics and Insurance Advisory Committee,) or by government taking advice from specialist bodies already active in the field (such as the British Society for Human Genetics, the Royal College of Pathologists or the Clinical Molecular Genetics Society⁵). The list of tests under such regulation would however require to be under continuous review: a genetic variant previously thought only to be important in the way a drug is metabolised by the body might turn out to be a predictor of a common serious disease such as cancer.

With appropriate consent, should results from direct genetic testing be recorded in the GP record? Should this be encouraged within any future regulatory system? (Q13)

This would require explicit consent. While it is recognised that genetic information is predictive (to a variable extent -see above) of future healthcare needs, one of the reasons why 'direct to consumer' tests may be popular is to bypass the GP and therefore keep the result from the medical record. Recent thinking in medical informatics is certainly to try and join up disparate information systems but it is vitally important that this does not disempower individual patients. Some patients may not trust the security and confidentiality of medical records, fearing that they may leak to employers and insurers. In these cases the option of extra-NHS testing may be appealing. Results from non-regulated tests are unlikely to be sufficiently predictive to justify incorporation into health records.

What are your views on the arrangements for retaining and using samples and data (Q14)

We support the recommendation, given at section 2 (page 9) of the current code, on confidentiality and security and the need for samples to be destroyed after a short time unless prior consent has been obtained for secondary use. However it should be noted that mistakes in testing might not come to light immediately. For example, a situation has recently arisen at a (NHS-funded) molecular genetics testing centre in Scotland where it appears that samples were subjected to the wrong test and consequently individuals were mistakenly told they were not at high risk of developing neuro-

⁵ See <http://www.bshg.org.uk/>, <http://www.rcpath.org/> or <http://www.cmgs.org/>

degenerative conditions. Intensive retesting is now underway but has been complicated in some cases by a need to obtain fresh samples from some individuals.

'Right to know' versus 'need to be protected from information one is not prepared for'

The arguments for and against 'paternalism in the age of consumerism' are well laid out at paragraph 44 of the consultation document. The right to information about oneself not available through a test on the NHS is not compromised by the regulation we propose if provision through a private medical provider is allowed. Careful thought will need to be given to the potentially conflicting issue of how to protect relatives of testee. We cannot see an immediate solution to this latter point.

Requirement for counselling to be made available by the test vendor

If the test is for a genetic mutation known to exist in a family member then the consequence may be well enough understood for counselling not to be necessary. In such a case it would seem unfair to expect someone to pay for counselling that they don't need. Alternatively, and particularly in regard to cancer genetics for example, there may be considerable need to have risk ratios set in context. We would suggest that complex tests involving family history are not made available without registered health professional interaction but that where pharmacogenetic (genetic influence on efficacy of drugs) or nutrigenetic (dietary advice based on genetic profiling) are offered that counselling options are available for those who want them. Minimally information indicating the limitations of the approach and the context of any risk ratios provided must be given.

Impact of extra-NHS testing on primary care health workers

Even if counselling is offered by the vendor with tests, and taken up, there does seem to be considerable potential for testees who have received complex information to be tempted to grab some GP or other practice team member's time to have it explained. This requirement can be partially ameliorated by requiring tests for family susceptibilities to be done through a medical practitioner. However a patient clutching a printout that claims to describe what their genetic profile argues they need to do with their life, in terms of diet and other lifestyle changes, may also require considerable time for interpretation and advice. In either case there will be enormous consequences for training: understanding of genetics, its limitations and its consequences is far from universal in today's NHS.

Consequences for specialist genetic service providers of extra-NHS testing

Extra-NHS testing will also have consequences for specialist NHS genetics services, in terms of time for follow-up, repeats of tests and specialist counselling. However this may not be preventable by sensitive regulation: a consultant geneticist contacted during preparation of this response replied gloomily that 'it seems like more work for us regardless of what the decision is'.

Advertising (Q2)

We note the controls on advertising described at sections 19-21 of the consultation document and in particular the point in para 19 about legislation and codes relating to promotion of prescription medications. Perhaps the issue of advertising tests is less important if access to the actual tests (where they are judged to be sufficiently predictive) is controlled. This would reflect the situation with promotion of prescription medications in the US.