Responses to Nuffield Council consultation on genomic medicine

I have answered selected questions in which I have the greatest expertise; in addition I have added (under Q15) a paragraph on a question which the consultation document does not cover, the development of large-scale and valuable biobanks by direct-to-consumer genomic testing companies, which I view as their main motivation for offering cut-rate genomic profiling.

Q1 Health care as a consumer good

I think this development is undesirable, as part of the growing commercialisation of the human body and of medicine. Biotechnology firms perceive very large markets in such areas as umbilical cord blood banking and direct-to-consumer (DTC) genetic testing, whereas traditionally they directed their energies at 'niche' markets associated with particular diseases. The 'worried well' of rich, healthy people who can afford DTC genetic testing are really the ideal market, one which companies are tapping as part of a general shift towards 'pre-symptomatic' treatment by the drug and healthcare industries.

With the expiry of patents on many successful medicines, and with the move into biotechnology by some companies with no previous holdings in this area, firms are casting about for new 'products': not in itself an undesirable development, but ominous if there are contravening arguments that these products risk harming vulnerable groups. In the case of umbilical cord blood, the risks of harm to mothers and babies have been documented by the Royal Collection of Obstetricians and Gynaecologists in its 2006 report on cord blood banking. These potential harms led the College to recommend that cord blood should not be banked routinely; in addition to these professional guidelines, private cord blood banks must be licensed by the Human Tissue Authority.

There are few such protections in the case of DTC genetic testing, except insofar as the kits themselves are covered by the MHRA. It is helpful that the Lords committee on genomic medicine has recommended that the level of risk classification should be increased from low to medium, but otherwise they propose to continue a light-touch regulatory regime inconsistent with that in many other European countries and US states. France, Austria, Switzerland and Germany ban direct-to-consumer genetic testing altogether, as do roughly half of American states. The Human Genetics Commission is now developing a code of practice, which the DTC industry actually does not oppose, but the question seems to be who will enforce it, as there appears to be little appetite in government for doing so.

Q2 Validity of information

Consumers who have paid for DTC genetic tests are more likely to think they should believe the results, on a cognitive dissonance basis: if they've invested so much money in them, the tests must be accurate. The same doesn't apply to what one reads casually in a newspaper or magazine. Companies' advertising and websites also appeal strongly to people's sense of 'empowerment' and 'individuality', making DNA testing seem innocent 'fun' like genealogy services, so that customers don't necessarily think of their wares in the same way that they would a health-related newspaper article. Additionally, the websites often 'blind with science', including articles and recent research findings. There's nothing wrong with giving the consumer scientific information, of course, provided that it really does represent the evidence base, which is not always the case for many of the commonest conditions such as cancers and heart disease. Professor Andrew Wilkie of the University of Oxford recently stated at a conference on DTC testing that for most tests, the SNP association has not been robustly demonstrated. In some instances where the link is clear, for example between the BRCA1/2 genes and breast/ovarian cancer, patent restrictions deter all but one of the DTC companies from offering tests. (The exception is 23andMe, which has decided to flout the Myriad Genetics patent.)

Q4 Who pays?

This is a major question. Similar issues arise with 'reproductive tourism' or organ transplants paid for abroad, where the NHS has to 'pick up the pieces' if things go wrong, but the breadth of the potential DTC genetic testing market is very much greater. I would favour the solution proposed of levying a charge on private providers of DNA profiling services, although there will be strong objections from companies based outside the UK, which is the majority of firms. However, if they want access to UK audiences, then it seems fair that they should pay a levy on every test performed on a UK resident. (Given that other European countries and many populous US states like California are denying them access altogether, they will probably still be glad of the UK audience, even if they have to pay a levy.) I don't think it will be feasible to charge patients themselves for follow-up diagnosis and treatment without undermining the principle that the NHS is free at the point of access.

Q12 Regulation

DNA testing services should have to satisfy a higher level of regulation in the private sector than in the NHS, where patients are protected to a much greater extent by the GP as gatekeeper, by professional guidelines and by negligence actions as an ultimate option in medical law. It is wrong that the situation is now the reverse, and in fact there are strong arguments, accepted even in free-market countries like the US, in favour of regulation extending to a total ban. This is an area where market failure is inevitable: there is an imbalance of information between consumer and provider, and no reason for consumers to

suspect that the limited information they do have might not be complete and true, particularly in an area where consumers have such inflated, media-fuelled expectations of the science as genetics.

A survey by Which in 2009 revealed that 79% of consumers agree that DTC genetic tests should be strictly regulated. What people actually want is regulation, which turns the usual argument about paternalism on its head. If it is paternalistic to deny people what they want, then the genuinely paternalistic course is to allow unregulated genetic testing, not to prevent people from getting unregulated genetic tests on spurious grounds of individuals 'right to know'.

Pregnancy tests are a very faulty parallel, since they provide a definite 'yes' or 'no' answer. By contrast, DTC genetic tests are probabilistic in their results, making the risks very hard to understand, and the probabilities will change every time a new genetic association is discovered. The specificity and sensitivity of most DTC tests is poor, and genomic medicine is still unable to account for more than about six to ten per cent of the variance in susceptibility to common diseases (as opposed to rare single-factor genetic conditions like Huntington's disease). 'Yes' or 'no' just isn't on the cards, but people may well assume from common consumerised tests with which they're familiar, like pregnancy tests, that they can in fact know for certain. In addition to implying that more certainty is possible than the science will support, there are allegations that DTC firms 'cherry-pick' favourable results.

Q15 Other issues not covered by consultation document

Many DTC firms, often with large initial capitalisation from other activities (e.g. 23andMe and Google), also hope to develop very large biobanks and databases which can be sold on to pharmaceutical companies and other businesses. Customers are frequently required to waive any onward rights in their data. The 'privacy statement' from 23andMe, for example, states that:

We] may allow an commercial research organization access to our databases …so that… the organization can search, without knowing the identities of the individuals involved, for the correlation between presence of a particular genetic variation and a particular health condition or trait. We may receive compensation from these research partners.

Some US commentators are convinced that this is where the money lies for the DTC companies, but that issue rarely arises in public debate outside the US, just as it is not covered by this consultation document. There are substantial issues about anonymisation of data and protection of consumers if these databases are being sold on uncoded to pharmaceutical companies for direct mail-shots. Even if the data are anonymised, consumers should be given the right to 'opt out' at any stage.

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.