Nuffield Council on Bioethics
28 Bedford Square
London WC1B 3JS

20 July 2009

Dear Sir/Madam,

Medical profiling and online medicine: the ethics of 'personalised' healthcare in a consumer age – consultation paper:
Comments from Breakthrough Breast Cancer

Breakthrough Breast Cancer is the UK's leading charity committed to fighting breast cancer through research, campaigning and education, and has established the UK's first dedicated breast cancer research centre, together with three new research units, in order to obtain our vision – a future free from the fear of breast cancer. Breakthrough campaigns for policies that support breast cancer research and better services, as well as promoting breast cancer education and awareness amongst the general public, policy makers, healthcare professionals and the media.

This submission reflects the views of Breakthrough, based on our experience of working with people with personal experience of, or who are concerned about, breast cancer. We regularly consult with members of our Campaigns & Advocacy Network (Breakthrough CAN) for their views on a range of breast cancer issues. Originally founded by women with personal experience of breast cancer, Breakthrough CAN brings together over 1100 individuals, regional groups and national organisations to campaign for improvements in breast cancer research, treatments and services. Through supporting and training members to become patient advocates in their own right, Breakthrough CAN aims to increase the influence of patients in decisions regarding breast cancer issues.

Breakthrough CAN members, health care professionals and researchers with an interest in breast cancer were consulted on the topic of electronic health records in February 2007. In addition, in 2007, Breakthrough spoke to members of its Genetics Reference Group (GRG) about their views on direct-to-consumer DNA profiling. The GRG are a group of over 170 people who have, or are interested in, a family history of breast cancer. Views and quotes from both of these consultations are included in this submission.

Breakthrough welcomes the opportunity to comment on *Medical profiling and online medicine: the ethics of 'personalised' healthcare in a consumer age*. Our comments are structured under the headings provided in the document.

Executive Summary

DNA profiling

An increasing number of DNA profiling services are becoming available direct-to-consumer, that is, without a medical intermediary. Breakthrough has concerns about the availability of such tests. First, our current understanding of how genetic variation contributes to disease risk is insufficient to support accurate risk assessment based on the results of such profiling. Second, the profiling is generally offered without associated genetic counselling. Third, there is a significant regulatory gap regarding DNA profiling offered outside of the NHS. In addition, as health claims made on websites are not classified as advertising, there is currently a lack of regulation on the claims companies can make for their tests. Finally, there is as yet no apparent clinical benefit associated with offering such tests to the general public.

For all these reasons, Breakthrough believes that all genetic or DNA profiling tests offered to patients in the UK should be thoroughly evaluated before coming to market. Once tests have been accepted for use in the UK, they should be offered only through a medical intermediary together with genetic counselling and should not be advertised directly to the public.

Any information provided online about DNA profiling tests should be comprehensive and accurate. Companies should be encouraged to apply for certification to indicate that they have adhered to ethical standards when presenting information and that the source and purpose of data on websites is clear to readers.

To ensure that patients receive unbiased genetic counselling from a properly trained health professional, it would also be beneficial to develop a mandatory regulatory code to govern the provision of genetic counselling.

<u>Online diagnostic information</u>
Organisations offering online diagnostic information should be encouraged to apply for certification so that patients know where to look for clear and accurate information.

There is a need for more research into health information-seeking behaviour in order to assess the impact of the increasing availability of online diagnostic information.

<u>Electronic patient records</u>
It is important that if an electronic records system is developed, it must be of value for both individual health care and public health. Where records are being kept by a healthcare provider such as the NHS, systems and appropriate training should be in place to ensure that information held on electronic patient records is accurate, secure and up-to-date. Providers of such records should ensure that patients are not denied the right to decide who may access particularly sensitive information.

The use of anonymised electronic patient data in research is a valuable resource that presents no risk to patients. The use of selected identifiable data should also be permitted, in both circumstances where express consent has been obtained from the patient, and where despite every effort such consent cannot be obtained, but approval by ethics and other regulatory bodies has been granted.

<u>Telemedicine</u>

It is very important that all people in the UK have access to free and appropriate health care services regardless of where they live. The most appropriate method of providing care may vary depending on individual circumstances. In some areas and for some patients, telemedicine may be the most suitable option and trials should be considered.

Section 1: Introduction

Question 1:

1.1 The development of DNA profiling services follows recent increases in our understanding of the genetic variation underlying common complex diseases such as breast cancer. In future, this knowledge could revolutionise disease prevention and treatment, but there is concern that these discoveries are being prematurely used for DNA profiling.

Our understanding of how genetic variation contributes to disease risk is still limited. Considerable research is still needed, including further investigating the individual contributions of specific genetic changes to disease, as well as how these changes interact with each other and with lifestyle and environmental factors to affect risk.

DNA profiling based on an incomplete understanding of the risk associated with genetic changes has the potential to cause harm by leading to unnecessary concern or false reassurance for those taking the test. For example, as our knowledge of the genes associated with breast cancer is incomplete, there is a concern that the current genetic tests which look at changes in genes other than the known breast cancer genes (BRCA1, BRCA2 and TP53) will provide misleading information about breast cancer risk. This could lead to women who are told they are a "low risk" making decisions that negatively impact their health (such as not attending breast screening appointments). It could also lead to unnecessary anxiety in women who receive a "high risk" result. People who have taken a direct-to-consumer test may be driven back to the NHS to find out more about their results, potentially putting primary healthcare services under pressure.

"What would this test do to GP time if women were walking in having bought a test on the internet which told them they were at risk and the time being used managing their fears unnecessarily… it might make GPs a bit exasperated so when they do get a genuine case they might miss it."
~GRG member

1.2 The risk of the potentially harmful effects of direct-to-consumer DNA profiling is increased due to the lack of appropriate genetic counselling services associated with them. Genetic counsellors are trained to guide people through the difficult decisions of whether to undergo genetic testing, the implications of their results for themselves and their families and of the choices that are available to them to manage their risk. Therefore, Breakthrough believes that genetic testing should only be offered as part of a medical consultation with an appropriately trained healthcare professional so

that any results can be explained, medical options communicated and support and advice provided.

1.3 Another concern is the significant regulatory gap relating to genetic testing offered outside the NHS. Within the NHS, the UK Genetics Testing Network (UKGTN) evaluates the effectiveness of genetic tests before they are adopted[1]. The UKGTN assesses tests according to clinical and analytic validity as well as clinical utility.[2] However, Breakthrough is not aware of any body with responsibility for evaluating genetic tests or DNA profiling offered outside the NHS. There is currently no mechanism to determine whether such tests are clinically relevant and companies are not prevented from making unsubstantiated claims about the usefulness of their tests for predicting health outcomes. Companies providing genetic profiling have no obligation to produce tests which are based on good clinical evidence of health benefit. There is therefore a need to develop an evaluation and regulation system to assess all genetic tests for clinical validity, analytic validity and clinical utility before they are permitted to reach the UK market. This would help to ensure patient safety and confidence in new technologies.

1.4 Finally, at present there is little apparent benefit in offering DNA profiling to predict breast cancer risk in the general population. We do not yet know how lifestyle factors can alter breast cancer risk associated with these genetic changes. For those likely to be at high risk of developing breast cancer, appropriate, clinically useful DNA testing (for faults in BRCA1, BRCA2 and/or TP53), counselling, early detection techniques and risk-reducing options are already available. It is questionable how offering DNA profiling, looking at genetic variation we do not fully understand, offered without counselling and without any proven interventions to reduce risk, would add value to services already available.

Question 2:

2.1 As detailed in section 1.3, Breakthrough believes that DNA profiling services are currently under-regulated. A lack of regulation on genetic tests offered outside the NHS currently allows tests which have little analytic validity, clinical validity and clinical utility to come to market in the UK.

2.2 An additional concern is that the majority of direct-to-consumer genetic tests are available via the internet. As health claims made on websites are not classified as advertising, there is currently a lack of regulation on the claims companies can make for their tests. This means that commercial companies may overstate the role of genes in common complex diseases and the ability of their tests to predict risk, resulting in the public receiving misleading health information. As detailed in section 1.1, this could lead to patients making

[1] http://www.ukgtn.nhs.uk/

[2] Clinical validity refers to the relationship between the genetic change being tested for and the disease or condition, analytic validity refers to the accuracy of the test in identifying the genetic change and clinical utility means the likelihood that the test will lead to an improved clinical outcome.

health decisions that could potentially harm their health. Therefore, stricter restrictions on the advertising of genetic tests should be considered.

2.3 As discussed in section 1.2, DNA profiling services offered direct-to-consumer are generally provided without associated genetic counselling. In the absence of a medical professional to provide information and support, it is extremely important that any information provided by the company has a high degree of scientific accuracy, and is thorough, clear and balanced. Detailed information, in plain English, should be made available to those ordering and taking the test to explain the ability of the test to predict disease risk, its accuracy and its clinical relevance.

While accuracy of information may be difficult to regulate, companies could be encouraged to apply for certification under the new Information Standard.[3] Under this Standard, an organisation's processes for developing information will be assessed, for example its process for making sure information is consistent with the latest clinical evidence. Organisations that attain the Information Standard will be entitled to display a quality mark on their information materials This should help consumers to easily determine whether information is trustworthy.

Similarly, the Health on the Net Foundation offers a certification to indicate that web site developers have adhered to ethical standards when presenting information and that the source and purpose of data on websites is clear to readers.[4]

Members of the GRG told us in 2007 that clear information is of great importance when considering genetic testing:

"It is essential to have clear written information to go with any testing. Even with counselling, it is impossible to absorb all the information in one go."
~GRG member

Question 3:

3.1 As discussed in section 1.1, current DNA profiling services are based on an incomplete understanding of the role of genetic variation in disease.

3.2 As our knowledge develops, it may be possible that accurate and clinically useful DNA profiling could be offered to determine disease risk. Under these circumstances, people might be encouraged to have DNA profiling in order to determine their disease risk and help them to make choices about appropriate interventions to manage their risk.

[3] Department of Health. *The Information Standard: A new certification scheme for Health and Social Care Information. Questions and Answers.* May 2009.
[4] http://www.hon.ch/

3.3 The choice of whether or when to undergo genetic testing, including DNA profiling, should always rest with the individual. Each patient would require appropriate genetic counselling in order to make an informed decision. If DNA profiling were adopted as a common measure for the general population, this would have implications for the genetic counsellor workforce.

Section 2: Electronic health records

Question 5:

5.1 Breakthrough broadly welcomes the development of any system that will enhance breast cancer services and treatment, improve the breast cancer patient experience and augment breast cancer research. It is important that if an electronic records system is developed, it must be of value for both individual health care and public health. However, providers of such records should ensure that patients are not denied the right to decide who may access particularly sensitive information.

"*For the most part I would support the notion of electronic patient records in that [they] provide (a) greater continuity of care with all members of the multidisciplinary care team able to communicate with each other and have instant access to decisions made by other carers regarding individual patients. This is particularly important when hospitals are split over several sites, or where radiotherapy or chemotherapy is held at a regional centre but the local unit is responsible for local care; (b) it avoids the issue of notes getting lost in transit between sites.*"
~ Consultant breast care nurse.

"*If a patient has medical conditions or treatments that [they] do not want others to know about they should be able to have that information kept private and not kept on the electronic record*."
~ Breakthrough CAN member.

5.2 Where records are being kept by a healthcare provider such as the NHS, systems should be in place to ensure that information held on electronic patent records is both accurate and up-to-date. Such accuracy is essential to both safe and effective treatment, and for the validity of any research based on the information stored.

"…when patients are being treated in hospital the experts need all the necessary information to give the best diagnosis and treatment. Ideally 100% security of records is needed. [I] hope that electronic records lead to more accurate and efficient diagnosis, treatment and research." ~ Breakthrough CAN member.

5.3 All staff responsible for filling in or maintaining electronic patient records must be fully trained in data security and patient confidentiality. It is also important that all and any access to data takes place only in specified circumstances. Only healthcare professionals directly responsible for a patient's care should have access to the full contents of their electronic records, with support staff and other individuals only being granted access appropriate to their role.

Any electronic records system established should be protected by the most thorough and up-to-date security hardware and software available, accompanied by effective systems to both limit the damage caused by and inform patients of any breaches.

"*GPs and hospital doctors only [should have access]. Not insurance companies, police (without court order), social services or any other party without express consent."*
~ GP.

"*No relatives, friends or acquaintances of patients should have access without prior consent by the patient."*
~ Breakthrough CAN member,

"*There needs to be prevention, however, of outside individuals walking up to hospital computers and being able to look at other people's records."*
~ Epidemiologist .

5.4 The use of anonymised electronic patient data in research is a valuable resource that presents no risk to patients. The use of selected identifiable data should also be permitted, in both circumstances where express consent has been obtained from the patient, and where despite every effort such consent cannot be obtained, but approval by ethics and other regulatory bodies has been granted.

"*…as far a[s] cancer is concerned all research is good and therefore information should be available for medical research… without the patient being identified."*
~ Breakthrough CAN member,

"If active consent was required, then large scale research would become very difficult, and potentially selected and hence biased, which would seriously damage the validity and hence value of doing the studies... For identifiable data, I think ethics committees should decide whether there could potentially be harm, and whether, on a case-by-case basis, individual consent is required."

~ Epidemiologist.

Section 3: Online health information

Question 6:

6.1 There are both risks and benefits of providing diagnostic information online. Online diagnostic information may under certain circumstances be helpful, for example to reassure the "worried well" who are seeking health information and advice. However, for others it may cause concern and lead to unfounded fears of disease. Similarly, for those who are ill such software may help encourage them to seek medical advice, but could also serve to falsely

reassure them. Much depends on the accuracy of the information provided and the quality of advice offered, such as signposting to relevant information. Organisations offering online diagnostic information should be encouraged to apply for certification under the Information Standard (see section 2.3) so that patients know where to look for clear and accurate information.

6.2 We do not yet have a good understanding of health information-seeking behaviour and therefore it is difficult to assess the impact of the increasing availability of online diagnostic information. There is a need for research to address the following questions: Under what circumstances do people access diagnostic information? What is it that they wish to find out? How do they use the information? Does finding out the information make them more or less likely to seek medical advice? Does the availability of such information reduce or increase the impact on health service capacity?

Section 5: Telemedicine

Question 10:

10.1 It is very important that all people in the UK have access to free and appropriate health care services regardless of where they live. The most appropriate method of providing care may vary depending on individual circumstances. Decisions need to be based on a balance between cost and the needs of individual patients. This should include consideration of how mobile the patient is, what transport is available to them, the patient's knowledge of and ability to use any technology required for telemedicine, the relative costs of telemedicine versus transporting the patient or health care professional, and whether there is a medical need to see the patient in person (for example whether a physical exam or medical tests will need to be carried out in a clinic). In some areas and for some patients, telemedicine may be the most suitable option.

10.2 For breast cancer, telemedicine may be more or less appropriate depending on the stage of care. For example, initial diagnosis will need to be carried out in the clinic due to the need for specialised equipment and physical examination, whereas certain aspects of follow-up care might under some circumstances be effectively carried out via telemedicine.

10.3 Trials are needed to determine the effectiveness and appropriateness of telemedicine as opposed to other alternatives. For example, NHS Orkney in Scotland has been testing methods including telemedicine for delivering more cancer services and treatments in local communities.[5] Such studies should be

[5] Scottish Government (2008). *Better cancer care, an action plan,* p57.

followed up to gain an increased understanding of the risks and benefits of telemedicine.

Section 8: Body imaging and DNA profiling services: cross-cutting issues

Question 12:

12.1 How genetic counselling is provided, and by whom, is of the utmost importance to ensure that the patient receives unbiased and useful information. As advocated by the Genetic Counsellor Statutory Regulation Steering Group, it would be beneficial to develop a mandatory regulatory code to govern the provision of genetic counselling.[6]

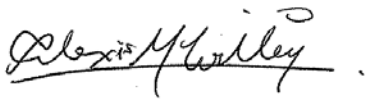
Question 14:

14.1 A better understanding of the genetic variation underlying disease could revolutionise the way we think about disease prevention and treatment. Therefore, funding bodies should continue to invest in genomic research.

14.2 However, until such a time as our understanding of this genetic variation and its interaction with lifestyle and environmental factors improves, there is a risk that DNA profiling could cause unnecessary concern or falsely reassure those taking the test that they are not at risk of disease, (see section 1.1.) There is an urgent need for improved regulation of DNA profiling tests offered outside the NHS to ensure that only clinically useful tests, associated with appropriate genetic counselling and comprehensive, accurate information about the risks and benefits are allowed to reach the market in the UK, (see section 1.3.).

Thank you for the opportunity to respond to this consultation on medical profiling and online medicine. If you have any questions or wish to discuss any of the comments made in this submission in more detail, please contact Dr Caitlin Palframan, Senior Policy Officer (Genetics), on 020 7025 2469 or caitlinp@breakthrough.org.uk.

Yours sincerely,



Alexis Willett
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[6] http://www.agnc.org.uk/About%20us/GCSRSG.htm