

## **Breakthrough Breast Cancer memorandum to the House of Lords Science and Technology Committee Inquiry into Genomic Medicine**

### **1. Executive summary**

- 1.1 Breakthrough Breast Cancer welcomes the House of Lords Science and Technology Committee's inquiry into genomic medicine. This is a rapidly progressing area of research and a new framework for strategic development is necessary to ensure that patients can continue to benefit from advances in genetic and genomic research.
- 1.2 Funding priorities should be identified in order to support the rapid scientific advancement of genomics. It is important that the Government and other funding bodies are able to provide medical researchers access to new technologies. Investment in translational research is essential so that advances in basic research can benefit patients in the clinic.
- 1.3 The generation of large amounts of data from next-generation technologies that allow for rapid and relatively inexpensive genome sequencing will pose significant challenges. The format of this data is important for its use and translation and there is a need for standardised annotation of accompanying clinical and pathology data. It will also be important to ensure that there is an appropriate balance between access to genomic data and security of patient or personal data.
- 1.4 Research to identify biomarkers that will assist in diagnosis and the specific tailoring of treatment regimens should be encouraged and assisted.
- 1.5 Whole genome association studies may lead to a better understanding of the genetic variation underlying disease and could revolutionise disease prevention and treatment. However, these discoveries should not be prematurely used for genetic testing.
- 1.6 There is a significant regulatory gap relating to genetic testing offered outside the NHS. It is important that an appropriate regulatory framework is developed in this area to ensure patient safety and confidence in new technologies.
- 1.7 Genetic testing should only be offered as part of a medical consultation with an appropriately trained healthcare professional. It would be beneficial to develop a mandatory regulatory code to govern the provision of genetic counselling.
- 1.8 Continued funding for improvements in genetics services is necessary. The continued supply of genetics counsellors and education of primary healthcare professionals about genetics and risk must be ensured.
- 1.9 People should be protected from genetic discrimination in insurance and employment. The Moratorium which currently prevents insurers from using the results of predictive genetic tests to set premiums for life, critical illness and income protection insurance has recently been extended to 2014. It is important that the Moratorium continues to be extended in future or that it is replaced with legislation which gives at least the current level of protection, if not more.

### **2. Introduction**

- 2.1. Breakthrough Breast Cancer is the UK's leading breast cancer charity committed to fighting breast cancer through research, campaigning and education. Breakthrough has established the UK's first dedicated breast cancer research centre, in order to achieve our vision: a future free from the fear of breast cancer. Breakthrough campaigns for policies that support breast cancer research and improved services, as

well as promoting breast cancer education and awareness amongst the general public, policy makers, healthcare professionals and the media.

- 2.2. Breakthrough held its inaugural Genetics Summit in 2006 which brought together Department of Health ministers with some of the UK's experts in genetics research and NHS services as well as women with a family history of breast cancer. A briefing outlining discussions held at the Summit is included as Appendix A. Breakthrough will be holding its second Genetics Summit in 2008.
- 2.3 Breakthrough works closely with healthcare professionals, patient advocates and researchers. Our memorandum reflects the views of Breakthrough and members of its Genetics Reference Group (GRG) – which is made up of over 100 people who have, or are interested in, a family history of breast cancer. GRG members play a vital role in informing Breakthrough on a wide range of issues, ensuring that our genetics and family history policy, campaigning and education activities continue to reflect the views of people with a family history of breast cancer. Members campaign locally and nationally on issues that affect people with a family history of breast cancer, such as reducing waiting times for genetic test results and the provision of local family history services. Breakthrough has also consulted healthcare professionals and breast cancer researchers in the development of this memorandum and the views of these stakeholders are reflected here. **Breakthrough staff and members of our Genetics Reference Group would be willing to provide oral evidence to this inquiry, if the committee would find this useful.**

### 3. Policy Framework

- 3.1 In 2003 the Government's policy on human genetics was set out in its Genetics White Paper *Our inheritance, our future: realising the potential of genetics in the NHS*, which identified where investment and support was needed for the NHS to better and more efficiently apply genetics advances to patient care<sup>1</sup>. It laid out strategic plans to invest in genetics services, research and development, to involve the public in developing the future role of genetics in the NHS and to ensure that public confidence in genetics was maintained.
- 3.2 The Genetics White Paper Review (2008) stated that more time was needed in order to embed many of the aims of the original White Paper in the NHS<sup>2</sup>. Therefore it will be important to continue to fund improvement in genetics services and research. A new framework for strategic development should be developed to ensure that patients can continue to benefit from advances in genetic and genomic research and it will be vital to continue to involve users of these services in the development of any such framework.
- 3.3 Within the NHS, the UK Genetics Testing Network (UKGTN) evaluates the effectiveness of genetic tests before they are adopted<sup>3</sup>. The UKGTN assesses tests according to clinical and analytic validity as well as clinical utility<sup>4</sup>. However, Breakthrough is not aware of any body with responsibility for evaluating either tests for biomarkers associated with disease susceptibility in the general population or pharmacogenetics testing<sup>5</sup> and there is a need to develop an evaluation and regulation system.

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<sup>1</sup> Department of Health. (2003.) *Our inheritance, our future: realising the potential of genetics in the NHS*.

<sup>2</sup> Department of Health. (2008.) *Our inheritance, our future: realising the potential of genetics in the NHS Progress Review*.

<sup>3</sup> <http://www.ukgt.nhs.uk/>

<sup>4</sup> 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.

<sup>5</sup> Pharmacogenetics testing examines genetic variation that may lead to a differing response to drugs between different patients.

- 3.4 The Human Genetics Commission (HGC) provides advice to the Government regarding the social, ethical and legal perspective on developments in human genetics. Recently, the HGC has expressed concern about a significant regulatory gap relating to genetic testing offered outside the NHS<sup>6</sup>. There is currently no mechanism to determine whether these tests are clinically relevant and companies are not prevented from making unsubstantiated claims about the usefulness of their tests for predicting health outcomes. Commercial companies producing these tests therefore have no obligation to produce tests which are based on good clinical evidence.
- 3.5 There is a need for a system to assess genetic tests for clinical validity, analytic validity and clinical utility before they are permitted to reach the UK market. If tests are proven to be useful and accurate this system would also help streamline the process by which they might be made available on the NHS. It would also help to prevent public trust in genetic technologies being undermined by the marketing of genetic tests of dubious value.
- 3.6 Two key principles must govern the development of any policy regarding the regulation of genetic tests. First, healthcare professionals and patients must be able to trust that the test they are using is accurate and capable of reliably predicting a given health outcome. Only useful and accurate tests should be permitted to reach the market. 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 and its clinical relevance. Second, genetic testing should be carried out only in combination with unbiased genetic counselling so that any results can be explained, medical options communicated and support and advice provided (see sections 8.2-8.3). The HGC has recommended that the recent *Organisation for Economic Co-operation and Development (OECD) Guidelines for Quality Assurance in Molecular Genetic Testing* and other international standards could serve as a basis for a code of practice for genetic testing that could govern all testing services<sup>7</sup>.

*“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.” Breakthrough Genetics Reference Group (GRG) member*

- 3.7 Stricter restrictions on the advertising of genetic tests should also be considered. The majority of direct-to-consumer genetic tests are available via the internet and, 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.
- 3.8 It is important to consider the harm that misunderstandings about the accuracy and usefulness of genetic tests could cause for people concerned about their risk of developing a disease. For example, people concerned about their risk of breast cancer could be put off from going for breast screening if they received a “low risk” result from non-comprehensive genetic testing, or might be caused undue distress if they received a “high risk” result without appropriate counselling. 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. As our knowledge of the genes associated with breast cancer is incomplete (see section 4.4), there is a concern that genetic tests looking at changes in genes other than the known breast cancer genes BRCA1, BRCA2 and TP53 will provide misleading information about breast cancer risk at this time.

*“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*

<sup>6</sup> Human Genetics Commission. (2007.) *More Genes Direct: A Report on developments in the availability, marketing and regulation of genetic tests supplied directly to the public.*

<sup>7</sup> Organisation for Economic Co-operation and Development. (2007.) *OECD Guidelines for Quality Assurance in Molecular Genetic Testing.*

*fears unnecessarily... it might make GPs a bit exasperated so when they do get a genuine case they might miss it." GRG member*

#### **4. Research and Scientific Development**

4.1 Advances in genomics over the past two decades have impacted on almost every aspect of medical research. The sequencing of the human genome, and the genomes of other organisms, now mean that the contribution of almost any gene to a disease process can potentially be assessed. Genomics plays a vital part in the study and understanding of cancer biology. For example, in cancer research, genomic approaches have led to the identification of a number of genes involved in the tumourigenic process (the formation of a tumour).

4.2 Two new developments have had a large impact on cancer research. The first is the use of genomics to refine the diagnosis and treatment of cancer. Scientists are beginning to understand the differences between tumours in different individuals at the molecular level. Genomics is at the centre of these efforts and it is hoped that genomic-based approaches will enable the better diagnosis of cancer, the better selection of appropriate treatment options and a better understanding of cancer biology that will lead to the development of novel therapeutics. The second is the development of novel technologies such as next-generation sequencing. These technologies enable high speed and high throughput DNA sequencing and should give a better understanding of the "normal" variability of the human genome, as well as the variations that lead to disease<sup>8</sup>.

4.3 Setting up these new techniques can be associated with high costs, so it is important to ensure that the Government and other funding bodies are able to provide medical researchers with access to these technologies. Costs in this field are large and escalating, and funders must be able to keep up with and provide access to the latest technologies, which have an increasingly short shelf-life as this technology rapidly evolves:

*"With the onset of next generation sequencing technologies... we will soon see how well the funding bodies can respond to enabling the majority of medical researchers access to a novel, yet expensive approach that has the potential to revolutionise medical research." Scientific researcher*

4.4 Research at the Breakthrough Toby Robins Breast Cancer Research Centre, at the Institute of Cancer Research, is a good example of the use of genetic approaches to cancer therapy development in the UK. Women who inherit faults in the breast cancer genes BRCA1 and BRCA2 have an up to 85% chance of developing breast cancer in their lifetimes. Genetic techniques were employed to investigate mechanisms that could be used to selectively kill BRCA-deficient cells whilst leaving cells with normal BRCA1 and BRCA2 unaffected. This led to the identification of molecules called PARP inhibitors as potential anti-cancer therapeutics. A Phase I clinical trial of PARP inhibitors in 2006/7 was very successful and two Phase II trials are now underway to investigate PARP inhibitors for women with breast or ovarian cancer linked to inheriting a faulty BRCA gene<sup>9</sup>.

4.5 Research is increasingly uncovering new information about genome variation and risk of common complex diseases such as breast cancer via whole genome association studies. A better understanding of the genetic variation underlying disease could revolutionise the way we think about disease prevention and treatment. However, it is important that these discoveries are not prematurely used for genetic testing. There remains considerable research to be done to enhance our understanding of

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<sup>8</sup> Mardis ER (2008) "The impact of next-generation sequencing technologies on genetics" *Trends Genet.* 24(3):133-41.

<sup>9</sup>[http://www.breakthrough.org.uk/research\\_website\\_2008/clinical\\_trials\\_clinical\\_researchers/clinical\\_trials/parp\\_inhibitor\\_trial/index.html](http://www.breakthrough.org.uk/research_website_2008/clinical_trials_clinical_researchers/clinical_trials/parp_inhibitor_trial/index.html)

risk before this should be considered, including 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. Genetic testing based on an incomplete understanding of the risk associated with genetic changes has the potential to do more harm than good by causing unnecessary concern or falsely reassuring those taking the test that they are not at risk of disease. There is currently little clinical utility in offering such tests as we do not know how lifestyle factors can raise or lower any risk associated with these genetic changes.

*"[I'm] not sure we should be testing for some of this without evidence it will produce a benefit to patients." Clinical researcher*

*"If you are going to offer a genetic test it MUST be scientifically valid... The offer should not make any spurious claims that could not be justified in front of a panel of scientific peers (in the same way that academic research is reviewed)." GRG member*

## 5. **Data use and interpretation**

- 5.1 There are a number of projects within Europe, such as *Ensembl*<sup>10</sup>, that are developing software systems to provide free access to annotated genomes. Although these systems are remarkable sources of genomic information, there are concerns within the scientific research community that the complexity of the data prevents many scientists from both accessing and understanding the information they contain:

*"I think that it is widely believed that the bulk of the information contained within genomic studies are not shared so that the scientific community as a whole can get the most value out of them. The data are complex, and this is difficult." Scientific researcher*

- 5.2 Furthermore, the generation of large datasets from next-generation sequencing technologies will pose significant challenges to the storage, sharing and management of such data.
- 5.3 Researchers have indicated that a common public database would be their preferred approach for the use and presentation of genomic data as it would allow transparency and accessibility to the largest scientific audience available. Experience from open source projects has indicated that this approach is also an effective way for the scientific community as a whole to find and fix errors, thus safe-guarding information quality. However, it will be important to ensure that there is an appropriate balance between public access and security of any associated patient or personal data. Extra safeguards to research could also be added to databases by only allowing access to scientific users with appropriate institutional agreements.
- 5.4 The format of genomic data is important for its consistent and accurate use and translation into other research applications. Standards exist for the quality, management, annotation and exchange of genomic data, such as those developed by the Microarray and Gene Expression Data (MGED) Society<sup>11</sup>. However, the associated clinical and pathological information recorded in publications and databases of genomic studies can be inconsistent in format and quality and there are no equivalent standards for the reporting of supporting clinical information. Inconsistencies in associated medical data prevent genomic information from being interpreted for clinical benefit:

*"The value of genomic data is multiplied many fold by the acquisition and recording of appropriate clinical information." Scientific researcher*

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<sup>10</sup> <http://www.ensembl.org/index.html>

<sup>11</sup> <http://www.mged.org/>

*“...a very poor link exists (at its best) between genomic information and clinical/pathology annotation, which severely compromises the advances that would lead to genomic medicine.” Scientific researcher*

Therefore a requirement exists for standardised annotation of clinical and pathology data, with prior anonymisation to maintain patient confidentiality, not only at the time of sample acquisition, but also during data submission for publication. This could be facilitated through the development of classification systems and controlled vocabularies.

- 5.5 In order to facilitate breast cancer research, Breakthrough scientists are collecting research data from teams within the Breakthrough Research Centre as well as published external sources in order to integrate knowledge of breast cells and their interactions into a single database. This work aims to develop a data integration platform to facilitate the analysis and mapping of a range of biological datasets, including genomic information. It is hoped that such informatics work will facilitate the detailed mapping of key pathways involved in breast cancer tumourigenesis (tumour development) and metastasis (the processes by which cancer spreads from where it originally developed), as well as the identification of novel drug targets and the development of new and better potential drug compounds.
- 5.6 The link between evaluating genomic data and addressing its utility must be made via translational research and requires the input of disciplines including basic research and genomics, bioinformatics (including mathematical analysis and statistics) and clinical practice. Breakthrough hopes that the promotion and advancement of genomic-related translational research will fall under the remit of the Office for Strategic Co-ordination of Health Research (OSCHR).
- 5.7 The Governing Genetic Databases Project<sup>12</sup> will investigate current practices for the collection, storage and use of DNA and information used for genetic research as well as the laws and governance provisions that apply to genetic databases. The project will eventually make recommendations for more appropriate and effective governance methods and models. Breakthrough hopes that the recommendations will be taken forward and will lead to the development of a uniform system for the governance of genetic databases throughout the UK, and possibly Europe. It will, however, be important that any governance changes are made with the full consultation of the scientific community and patient organisations to ensure that they protect patient confidentiality without hindering the advancement of scientific and clinical research.
- 5.8 There is, however, potential for abuse in the use and interpretation of genetic information. Women with a family history of breast cancer are extremely concerned that the results of genetic tests may be used against them by insurers or employers, and some believe there should be legislation to prevent this:

*“I feel that people may lose out in obtaining jobs or keeping their jobs due to employers being able to find out that a particular person has a genetic predisposition [for a disease]. If the legislation is not there this may... allow employers to pick and choose their workforce on medical grounds and maybe the longevity of their workforce and not on how good they are at their job.” GRG member*

*“I believe that people will be discriminated against, and will either only be able to obtain insurance at high premiums, or even will become uninsurable.” GRG member*

- 5.9 Women with a family history of breast cancer have told Breakthrough that they might decide not to undertake a predictive genetic test for breast cancer risk if they knew their results might be used by insurers now or in the future. This is a real concern, as the results of predictive genetic tests can help them to make important decisions

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<sup>12</sup> <http://www.ggd.org.uk/index.cfm?fuseaction=home.index>

about their health, such as deciding whether to undergo preventative surgery. The decision to take a genetic test can also impact on the lives and healthcare decisions of children and other relatives. A child of an individual with a fault in one of the BRCA genes has a 1 in 2 chance of inheriting that fault.

- 5.10 At present, the Moratorium between the Government and the Association of British Insurers (ABI) prevents insurers from using the results of predictive genetic tests in setting premiums for life, critical illness, and income protection insurance<sup>13</sup>. The Moratorium means that people with adverse genetic test results are still able to get reasonable levels of insurance coverage.
- 5.11 The Moratorium has recently been extended so that it expires in 2014<sup>14</sup>. The Moratorium should continue to be extended or legislation, possibly via the proposed Single Equality Bill, should be put in place to enshrine the Moratorium by banning the use of predictive genetic test results in setting premiums for life, critical illness, and income protection insurance. However, care is needed to ensure that any such legislation would give at least the current level of protection that the Moratorium provides, if not more. This would allow people to undergo predictive genetic tests without the fear that their test results could be used against them now or in the future.

## 6. Translation

- 6.1 The behaviour of a cancer is based on its genomic profile. As the identification of the genes involved in tumourigenesis increases, this should eventually lead to the ability to determine the clinically-relevant genomic profile of any individual tumour, allowing targeted, individualised treatment and a better prediction of disease outcome. One such application of genomic research that is already used in the clinic is HER2 testing of breast tumours to determine if they will respond to treatment with Herceptin<sup>15</sup>.
- 6.2 Clinicians play a leading role in translational research. Breakthrough supports the development of clinicians who wish to undertake initial training in breast cancer research or who want to further their research careers through our Avon Clinical Research Fellowships and the Breakthrough Clinical Researcher Programme. A focus of our research strategy is translational research, for example in the analysis of gene expression in oestrogen receptor positive breast tumours to determine mechanisms of resistance to hormone therapy.
- 6.3 Investment in translational research is essential so that advances in basic research can be of benefit to patients in the clinic:
- “Without investment we fall behind scientifically and commercially. You have to get experience and knowledge to progress.” Clinical researcher*
- 6.4 The UK is well-placed to lead in the area of translational research, especially because of the quality of our clinical (e.g. tissue) specimens and the associated patient data that can be accessed via the NHS (e.g. diagnosis, disease outcome). However, it is important that electronic patient data is consistently coded and linked so that genomic data can be included in a useful manner (see section 5).
- 6.5 As genomic science progresses, new and better ways to analyse tissues and individuals will be developed and so it is unlikely that any one-off programmes of investment in this area would be ‘future proof’. It is important that there is continued investment in this area to ensure that UK science can keep up with advances in

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<sup>13</sup> HM Government and the Association of British Insurers. (March 2005.) *Concordat and Moratorium on Genetics and Insurance*.

<sup>14</sup> Association of British Insurers. (13 June 2008.) *Insurance genetics moratorium extended to 2014*. Press release.

<sup>15</sup> As part of the testing procedure, breast tumours may be subject to a technique called fluorescence in situ hybridisation (FISH) to determine whether the HER2/neu gene has been amplified in the tumour cells. Amplification of the gene and over-expression of the HER2 protein indicates that the breast tumour is likely to respond to treatment with Herceptin.

genomic medicine. Bodies such as OSCHR will play important roles in directing funds to and promoting mechanisms of the translation of genomic advances into clinical benefit.

## **7. Biomarkers and Epidemiology**

- 7.1 Women affected by breast cancer tell Breakthrough that they would value research into rapid and more accurate diagnostic techniques that would point to the most appropriate and effective treatment for them. Information on clinical outcomes is also important to them. Research to better identify biomarkers that will assist in diagnosis and the specific tailoring of treatment regimens should be encouraged and assisted.
- 7.2 One sub-group in which the identification of biomarkers will play an important role is women with a strong family history of breast cancer in whom no known heritable genetic mutation has been identified. These women often undergo preventative surgery to remove their breasts and ovaries without knowing for certain whether they have inherited a gene fault that will make them more likely to develop breast cancer. Breakthrough hopes that the recent formation of the network of Experimental Cancer Medicine Centres<sup>16</sup> will allow a greater focus on translational biomarker research to progress within the UK and for the benefits to reach patients as quickly and effectively as possible.
- 7.3 Genomic data will have an impact on data emerging from biobank projects by providing the potential to investigate gene-environment interactions. Such research may improve the understanding of cancer aetiology (the understanding of the cause or origin of disease) and may possibly lead to personalised, more effective, cancer prevention. Furthermore, the blood samples from biobank projects can be used to identify a greater number of cancer predisposition genes.

## **8. Use of genomic information in a healthcare setting**

- 8.1 As stated above (sections 4 and 7), genomic information that is linked to patient data on diagnosis, pathology and disease outcome could potentially lead to a better understanding of tumourigenesis and gene-environment interactions, and therefore the aetiology of cancer. As highlighted in section 5, it is very important that genomic data can be consistently and accurately linked to good quality pathology data in order to aid disease classification and that clinical data is coded and standardised. These measures will help to ensure efficient translation on research advances into the clinic.
- 8.2 The decision to take a genetic test is extremely personal, complex and often difficult. It is very important that those considering genetic testing have access to genetics counsellors 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:

*“In my experience the interview with the genetic counsellor before deciding whether to take the test was much more valuable than any test could have been. She was able to work out that there was a high probability given my family history that I would get a positive result and talk to me about how prepared I was for that kind of knowledge and what I might do with it.” GRG member*

*“In my case I still feel that I would like to talk to someone who can look at my case and give me up to date advice on what my family’s risk is and what we could be doing about it.” GRG member*

*“Lack of knowledge and a positive result could mean panic and some hasty decisions being made. Some form of counselling is required so people are aware of the consequences of a positive result.” GRG member*

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<sup>16</sup> <http://www.ecmcnetwork.org.uk/>

- 8.3 The decision whether to take a genetic test should be based on the needs and wishes of the patient and the clinical utility of the test. How genetic counselling is provided, and by whom, is of the utmost importance. 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.<sup>17</sup>
- 8.4 The rapid pace of developments in genomic technologies and medicine means that many healthcare professionals will need additional education support to improve their understanding and integration of genomics, as well as transcriptomics and epigenetics<sup>18</sup>. It will also be very important to ensure the continued supply of trained genetics counsellors to the clinic and to educate primary healthcare professionals on genetics and risk so they can refer people to genetics services as appropriate.

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<sup>17</sup> <http://www.agnc.org.uk/About%20us/GCSRSG.htm>

<sup>18</sup> Transcriptomics is the study of the expression level of mRNAs ("transcripts") in cells and reflects the genes being actively expressed at any one time. Epigenetics is the study of stable alterations in gene expression that do not involve changes to the basic DNA sequence. In this way environmental effects can cause genes to be turned on or off.

## APPENDIX A

### **Extract from Breakthrough Breast Cancer's Genetics Summit 23<sup>rd</sup> November 2006**

#### **PRESENT**

Andy Burnham MP, Minister of State, Department of Health

Prof Alan Ashworth (Chair), Val Davison, Prof Diana Eccles, Prof Gareth Evans, Prof Peter Farndon, Chris Jacobs, Mary Kennedy, Alastair Kent, Dr James MacKay, Dr Christine Patch, Dr Sarah Rawlings, Su Stenhouse, Prof Richards Trembath, Dr Andrew Tutt, Jacquie Westwood, Louise Wilder

#### Observers:

Diana Paine (Department of Health), Alicia Ionannou (Department of Health)  
Vicki Nash (Breakthrough Breast Cancer), Imran Hussain (Breakthrough Breast Cancer), Sarah Etwell (Breakthrough Breast Cancer), Dr Norman Freshney (Breakthrough Breast Cancer)

#### **THE 'GENETICS REVOLUTION'**

The session began with a presentation by Prof. Alan Ashworth on a perspective of the genetics revolution. Key points that emerged from the ensuing discussion were:

**Information for information's sake:** The Minister posed the question of how useful and usable genetic information will be. The clear view of the meeting was that genetic information is increasingly useful and usable.

**Cost implications for the NHS:** The more we know about how genetics impacts on diseases, the better we can be at targeting services and treatments to those most at risk of developing diseases. Such investment in targeted areas and disinvestment in others would potentially save the NHS significant financial resources. An example was provided where a particular drug is given to a targeted audience, rather than blanket coverage such as chemotherapy. Another example was provided where those most at risk are screened more regularly using the most appropriate techniques.

**Susceptibility tests:** The number of susceptibility tests available will increase as more targeted drugs come on to the market. It was felt by some that this could impact on the workload of NICE and that a wider discussion around the future of drug pricing for the NHS is needed.

It was suggested that there needs to be a discussion on whether the NHS should pay for this type of test. A particular view was that susceptibility tests need to be recognised as being clinically important before being offered.

**Improved integration of systems:** There was strong consensus towards developing a system of genetics networks to aid co-ordination and share best practice of service development and configuration. It was argued that some genetics services will need to be reconfigured. There was a suggestion that the networks could be similar to cancer networks already in operation.

**Transition from specialist to mainstream services:** It was recognised that now is a vital time for genetic services. Although genetics services are specialist services, there is agreement that in the future they will be part of the mainstream NHS. It was felt decisions taken over the next months and years would determine how well this transition would happen.

The Minister explained he was aware of the issue of the transition period from specialist to mainstream services and that genetics services will be at the heart of a preventative NHS.

**Payment by results:** It was highlighted that services are currently commissioned differently across the country resulting in a difficulty to get a standard cost per episode. Therefore, careful work will be needed to ensure any tariff set under Payment By Results is adequate to meet the costs of providing the genetics service.

**The research agenda:** The Minister made clear he felt that research was moving up the Department of Health agenda, particularly as both the Comprehensive Spending Review and the Cooksey Review are taking place.