

HOUSE OF LORDS  
MINUTES OF EVIDENCE  
TAKEN BEFORE  
THE SELECT COMMITTEE ON SCIENCE AND TECHNOLOGY  
(SUB-COMMITTEE II)

**GENOMIC MEDICINE**

WEDNESDAY 9 JULY 2008

DR HELEN WALLACE and MR ALASTAIR KENT

PROFESSOR PETER FARNDON and MRS JACQUIE WESTWOOD

Evidence heard in Public

Questions 343 - 420

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WEDNESDAY 9 JULY 2008

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Present

Broers, L  
Colwyn, L  
Finlay of Llandaff, B  
Krebs, L  
Northesk, E of  
Patel, L (Chairman)  
Perry of Southwark, B  
Taverne, L  
Warner, L  
Winston, L

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**Memoranda submitted by GeneWatch UK and Genetic Interest Group**

**Examination of Witnesses**

Witnesses: **Dr Helen Wallace**, Executive Director, GeneWatch UK, and **Mr Alastair Kent**, Director, Genetic Interest Group, examined.

**Q343 Chairman:** Good morning. Welcome to all of you, particularly the members of public at the back. For you, there is information showing the declarations of interests of individual members if you wish to know that. Could I welcome our first witnesses: thank you for coming today and helping us with our inquiry. If either of you wish to make an opening statement before we go on to questions, I would like to hear anything you have to say but particularly how each of your organisations is funded. Who helps you with funding? Perhaps you might address that. When you speak for the first time, it would be helpful for the record if you introduce yourself and say which organisation you are from. Please feel free to make any opening statements you have and proceed with answering my question.

**Mr Kent:** Good morning. Thank you very much for the opportunity to present evidence. I am Alastair Kent. I am the Director of the Genetic Interest Group. The Genetic Interest Group is an alliance of approximately 140 charities and support organisations representing

and speaking on behalf of patients with the full spectrum of genetic diseases, from the very rare, single gene disorders to very common, complex conditions. In terms of our funding, we are funded from a diverse range of sources. We currently receive some money from our members in the form of subscriptions. We receive some money from the Department of Health through a section 64 grant; from the Medical Research Council; from a range of charitable trusts and foundations; and a proportion from industry, some of it from the pharmaceutical industry and some of it from other private sector organisations. Our policy is to remain independent of any one source of funding and not to accept money that is conditional on us arguing for any particular point of view or policy position. Our position comes from consultation with our members and from reflecting the views and opinions that come back to us on issues which are of current concern to them. We are particularly grateful to have the opportunity to present oral evidence to this Committee because this is clearly an issue of great concern to patients and families, largely because of the progress that we have seen in recent years, which has been astonishing in terms of the new knowledge of genetics and genomics that is emerging, but also we have to remember that we are at the beginning of a journey rather than at the end of it. In this context too, the long-term goal of producing effective available interventions for patients and families living with intractable diseases can often seem to be obscured by short-term excitements, and our hope is that this committee of inquiry will clearly separate the hope from the hype, the realism from the fluff, helping sustained, high quality biomedical research through the generation of a regulatory framework that is appropriate and proportionate, and which takes account of the risks and benefits to be gained from this work. That is probably all I should say at this point.

***Dr Wallace:*** Thank you for inviting me. I am the Director of GeneWatch UK, a small not-for-profit organisation. We basically take the view that the public should have a say about science and technology and how these new technologies are used and developed. We provide

public information and information about policy implications of genetics quite broadly. We are funded mainly by charitable foundations. We also have some funding from the European Commission's Science in Society programme and we get a small amount of funding from donations. Our key interest in this area is really the idea of expanding genetic testing or genome sequencing to the whole population on what the implications for society of those kinds of changes might be.

**Q344 Chairman:** Thank you very much. To pick up on the very thing you mentioned, we have had evidence about the myriad of tests that are already available. Whilst science is developing and these tests are being marketed and used, there does not seem to be any particular authority co-ordinating the whole activity. There is a myriad of different committees and organisations involved in giving advice either to professionals or to the public. What is your comment about that? Who do you think should be doing this?

**Dr Wallace:** I think there is a real gap in the current advisory system, so we certainly welcome some of the work done by the Human Genetics Commission and others. We are aware, for example, that there is a committee to assess the usefulness of these tests to the insurance industry, in terms of the Genetics and Insurance Committee, but there is no committee or regulator to assess the usefulness of the tests for health, so there is some assessment within the Health Service for tests offered there, but there is no routine system for analysing the clinical utility or validity of the tests. We have found that is of particular concern for the commercially marketed tests, where, as you know from our written evidence, there have been a large number of claims made which are not substantiated by the scientific evidence.

**Mr Kent:** It is important when looking at this to distinguish between what happens in relation to tests that are used in the NHS and those that are perhaps brought out in the private sector for use either in private medical treatment or for sale direct to the public. In the context of the

NHS, in our view the current UK Genetic Testing Network – and I know colleagues will be speaking to you later this morning about that – works well for the examination of utility of tests for single gene disorders and the model that the UK GTN has adopted could well be extended for the systematic evaluation of tests that are designed to address complex, multifactorial conditions. However, that would require an increase in the breadth of expertise and also in the resources available, to allow that mission to be accomplished efficiently and quickly, and not to put an undue delay into the transfer from research into clinical practice. In the private sector there is a much greater degree of confusion. There is an unwillingness of current potential regulators to get involved in looking at the claims that are made. The Advertising Standards Authority, the Trading Standards Authority, although potentially they have the power to regulate by looking at claims and such things, lack the competence. The NHRA, on the other hand, has the competence but seems to lack the authority, because its activities are regulated by the In Vitro Diagnostic Directive, the focus of which is simply: Does the test meet the claims that it says it does? Does it do what it says on the tin? That focuses on the technical adequacy, not the utility. In our view, there is a need for a body such as the NHRA, which does have the expertise, to be encouraged to extend its remit, not just to look at the technical accuracy of tests that are put on the market but also to put them into the context of whether or not they are useful, whether they give information, and the significance of the information that is provided. Unfortunately, genetic tests are generally deemed to be of low risk because they involve a cheek swab or a bloodspot, but the risk in the genetic test is not in the invasiveness of the procedure to take a sample but in terms of the potential impact of the information that is revealed by the analysis. That is where we would like to see the NHRA taking perhaps a higher profile in terms of looking at regulating these tests.

**Q345 Lord Krebs:** Perhaps I could pick up the theme of regulation. Mr Kent, in your introduction you mentioned the word “proportionality”, which is about balancing the

constraints on the industry in relation to research against the benefits. I wonder whether you could tell us how you would see balancing the need for regulation of genetic tests, particularly in terms of consent and confidentiality and privacy, against the potential benefits that might be conferred on family members or in terms of the development of research.

**Mr Kent:** Yes, certainly. I think it is important to distinguish between the clinical application of tests for use by family members and the research application. To take the clinical application first, clearly consent is a very important issue. In an ideal world, fully informed consent would be obtained in all cases. However, that is not always possible for a variety of reasons. In our view, the model that was created under the Human Tissue Act would potentially provide a way forward, although that is limited. The Human Tissue Act provides the opportunity, where consent is unclear or missing and it is not possible to go back to the originator of the sample, for the court to make a decision whether or not a particular intervention, a particular procedure, should be allowed. When the HTA was in front of Parliament, we argued that when consent had been withheld and there was no reasonable mechanism for going back to the originator of the sample to see if their withholding of consent still applied, the court could be given a similar power. The Government disagreed, feeling that that was contrary to the spirit of the Act. It seemed to us, however, that people change their minds, circumstances change, and what might have seemed inappropriate at one point could become appropriate and desirable at another, and if contact had been lost then the health benefits to be had by blood relatives should trump the absence of consent. With regard to research, in our view, identifiable samples that can be traced back to individuals or identifiable data that can be traced back to individuals should only be used in those instances where the identifiability is central to undertaking the research. A lot can be done with anonymous samples and sometimes that should be sufficient. However, with regard to the research uses in genetic information and genomic information, public surveys have shown

that, by and large, there is support in certainly patient groups and amongst the wider public for making sure that the best possible use is made of valuable data and samples. For example, when the Medical Research Council was examining the possible uses of the newborn bloodspot samples, which are potentially a hugely valuable resource for investigating complex disorders because they are an unselected population sample, a programme on public engagement did reveal widespread public support for research that was into interventions for serious diseases, for improving the technical and analytical quality of the service provided, and for also making sure that the information that was revealed was potentially fed back to those people who would be in a position to benefit from it in the fullness of time, when it was mature enough to be used for clinical applications.

***Dr Wallace:*** In relation to the regulation of the test in itself, the key issue really is an issue of trust and whether people can trust the information that they are being given. There is basically a consensus internationally around the kind of information that people would need to be assessed in order to do that as an established process, and that considers analytical validity and clinical validity and clinical utility. Obviously there are also ethical implications and social implications which would be very hard to regulate but, in terms of the kind of evidence that you need, I think the key is to distinguish between the right to know and the right to be misinformed or misled. We have to put in place something that prevents people from being given misleading or false information, and there are already established guidelines by the OECD in terms of what kind of information it would be best practice to provide. There is now a new protocol to the Council of Europe's Convention which states that there should be a requirement to meet certain levels of validity and utility, so it is very clear that it is not simply a question of wrapping red tape around the idea of providing these services; it is about meeting minimum and best practice requirements. In the case of research, I would say also that it is the issue of trust again that is important. Nobody wants to see research bound up in

red tape, but there are certain key issues that the public raised time and time again in consultations, particularly issues about intellectual property and conflicts of interest, for example, where there is often dispute amongst the experts about the best policy to have, and we would like to see those kinds of issues discussed very transparently at the start of the research projects and not brushed under the carpet.

**Q346 Lord Krebs:** Do you see the field of genetic testing as being fundamentally different from other areas of health benefit provision? Products that claim to improve health or improve your prospects of dealing with disease often make claims that are on the verge of validity, and the Advertising Standards Authority or the Trading Standards officers are there to make sure that companies do not tell lies, but do you see a fundamental difference between claims made by genetic testing companies and claims made by other healthcare companies?

**Dr Wallace:** I think the answer is yes and no. There are some aspects of claims that are made by other companies that would certainly benefit from more regulation and oversight. In general, a shift to more predictive testing, for example, would benefit from additional assessment of whether those predictions are valid. However, I think there are some special characteristics if you look at any area, and there are some special characteristics around genetic testing that we can identify. One is the difficulty of establishing utility. That requires different kinds of information than perhaps you tend to need for other biomarkers because we are talking about fixed risk factors. You are not advocating that the patient or the individual changes those risk factors; you are trying to tailor other products or advice to those risk factors, which is a more complicated thing to establish. The second is the broader picture issues. Genome sequencing, for example, is essentially a dual-use technology and it raises additional privacy concerns around the potential use for tracking individuals or their relatives. I am not saying there are not privacy issues with other types of information, but that does have a special characteristic in terms of being able to take a sample and identify an individual

on the database. Third, I would say there are some social and bigger picture issues around the fact that it is an emerging technology, where lots of companies are trying to do a lot of testing. There has been a lot of hype, so a lot of individuals think this is the future of medicine and they want to be at the forefront of getting those tests and, therefore, we have a social context where it is important that people can trust the information they are given.

**Q347 Lord Taverne:** In the paper that you submitted, which is a very valuable paper, you express a lot of concern about the corporate testing that is taking place. But there is a suggestion in the paper that you have reservations about the whole future benefit from genomic medicine in relation to common diseases to establish a relationship, in particular, to Huntington's chorea. Is that a general worry you have about the usefulness of genomic medicine, as such, in that context?

**Dr Wallace:** It is not a general worry about the usefulness of genomic medicine or the usefulness of research that could identify the role of genes in common diseases and therefore understand disease mechanisms, but it is a general worry about the future predictive value and utility of this type of testing. I think you will find if you talk to a lot of genetic epidemiologists that it is quite a widespread concern in that community. They very much still want to do the research – they think it would be useful to understand, for example, the mechanisms of appetite in obesity – but the claims that you would predict which individuals will become obese or will get heart disease and so on and be able to tailor treatments to those individuals do not seem to be borne out in general by the evidence. Of course, there are exceptions. There may be some useful tests, but the predictive value of that information is going to tend to be relatively small for these late onset complex diseases because of all the other factors that are involved.

**Q348 Lord Winston:** That is, Dr Wallace, most diseases.

*Dr Wallace:* Yes.

**Q349 Earl of Northesk:** Digging a bit deeper into the regulatory framework, in California, direct-to-consumer tests such as those offered by deCODEme and 23andMe have recently been banned if tests are carried out without medical advice or in unlicensed laboratories. Do you think there is a case for a similar ban under UK or EU legislation, or do you think that other approaches might be more appropriate and effective?

*Dr Wallace:* I think there is a case for a ban on offering tests directly to the public without medical information. However, I would prioritise the information requirements; for example, requiring accredited labs so that you know they are testing the claimed gene sequence and requiring that evidence of the interpretation of the test. Once you have that information, you might in the future well decide that some tests perhaps need less medical oversight than others, but until you have that information and until you know, for example, the potential for unexpected surprises, a gene that is linked with something that you were not expecting, then it is quite concerning that people can be offered those tests without really knowing the implications of the results before they take them.

*Mr Kent:* I take a different view. I do not think there is a case for a ban at the moment because that would make patients and families too dependent for their potential access to genetic information on the willingness of their doctor to prescribe that test for them. Given that there are significant constraints on NHS resources, given that physicians' knowledge about the potential utility or otherwise of genetic tests is variable, to put it at its mildest, that might in practice impose significant constraint on people's right of access to information which may be useful for them. Also, if there were to be a ban, there is a risk that you would simply create a black market of people operating from unregulated territories, and I would rather that people who wanted to supply tests to our members and their families were operating in a properly regulated and transparent environment. However, there is a case, I

think, for making test providers come clean about the limits and the possibilities of the tests they are trying to provide. They should put the evidence for their claims into the public domain in forms that are accessible to people, so that purchasers, whether they are non-specialist clinicians selecting a test on behalf of a patient or individual citizens buying directly for themselves, can make an informed decision about the quality of the test and the likely value of any information that might be revealed by it. I think there is also a case for a programme of awareness-raising amongst the public and amongst non-specialist professionals – and in that we must not forget the complementary practitioners, some of the nutritionists and what-have-you with alternative qualifications who are looking perhaps to genetic testing to advise on dietary modifications. We need to raise the awareness of the public and of the non-specialist professionals.

**Q350 Chairman:** Can you give an example, apart from genetic advice, of dietary advice that a nutritionist might give that might be linked directly to a gene?

**Mr Kent:** Yes. There was an attempt a couple of years ago now by a company called Sciona to market a programme of genetic tests that were claimed to be linked to an increased risk of developing a wide range of conditions. On the basis of the information that they claimed the analysis of the DNA revealed, they were offering dietary advice, saying, “Because you have this particular polymorphism you have an increased risk of that disease, therefore it would be sensible for you to include these sorts of things in your diet or to exclude those sorts or things from your diet.” A lot of it would amount to: “Eat five portions of fruit and vegetables a day – drink less alcohol – take more exercise – less fatty acids” but certainly there were claims being made about the predisposing risk that was revealed by genetic analysis and that that risk could be modified by targeted dietary interventions and modifications.

**Dr Wallace:** Could I add some examples, because we have investigated a number of these companies? One marketing technique that has been used is to market supplements associated

with the tests, so to sell the test via alternative healthcare providers who already have links with supplement companies and then to give advice on either standard supplements or in some cases special preparations and so on that are intended to reduce risk. That has happened in the UK with dietary-related conditions and certainly in the States it has also happened with things like attention deficit disorder in children. We are also aware of companies that are interested now in starting to market tests for psychiatric disorders like bipolar and schizophrenia and they might also start doing the same kind of thing, so that raises additional concerns about misinformation and the exploitation of potentially vulnerable people, including children.

**Chairman:** Thank you very much.

**Q351 Lord Colwyn:** Continuing with the theme of testing, we heard in evidence that in this country and in the United States – and in fact, Dr Wallace, you stressed this yourself earlier – it is essential to assess the clinical validity and clinical utility of emerging genetic tests for disease susceptibility. Who do you think should fund it and who do you think should carry out this research?

**Dr Wallace:** Again, I think it depends on the test and what it is proposed use is. There are some clear examples where we have a lot of evidence that a test may be useful; some of the pharmacogenetic tests like CYP2C9 for warfarin. That is publicly funded research now because we think it may well be useful in the Health Service to have that evidence. There are other situations, for example, lung cancer, where there is the idea of giving genetically tailored advice to smokers on who is at most genetic risk of lung cancer. That idea has a long history of funding from the tobacco industry. We know there is not a significant inherited component in lung cancer, we know it is likely to mislead individuals because there are many other smoking-related diseases, so it would be, in my view, wrong to put public money into that type of research. There is a whole range of conditions in between where a lot of existing

data will tell you now whether you have quite a good prospect of a good useful test that might merit some public money, or if a commercial company is proposing to do it let them pay for that research if you do not feel that is going to deliver significant public health benefit.

**Q352 Lord Colwyn:** Mr Kent, you are nodding. You agree with that.

*Mr Kent:* Yes, I broadly agree with that, the principle that if the private sector wants to develop a test then it should fund the research necessary in order to prove its utility and validity. The regulatory infrastructure, the capacity to evaluate that utility and validity should, at least in part, be funded publicly in order to guarantee its independence; however, there is no problem in my mind to charging a fee to those people who want to have their tests evaluated, providing there is a mechanism for allowing those tests which are perhaps commercially unattractive but which may have good clinical benefits through the system. If you look by analogy to the orphan medicinal products regulations, where therapies developed for rare conditions which are unlikely to be economically attractive to the private sector are able to get subsidies to help them through the regulatory process, in the forms of fee waivers and scientific advice and so on from the European Medicines Agency, providing there is that mechanism to protect the interests of those who may derive significant benefit from tests that would otherwise be unattractive to the private sector to develop, then the private sector pays for what it wants to do and that is a good principle.

**Q353 Baroness Finlay of Llandaff:** Could I follow up very briefly on the comment you made, Dr Wallace. Do you envisage tobacco companies potentially developing a “safe smoking” test to say that you are in a group that has a lower risk of malignancy based on your genes?

*Dr Wallace:* I have done some research that is not published yet based on internal tobacco industry documents and I know that they put a lot of funding into basically looking for lung

cancer susceptibility genes. The message they were putting out was that only one out of ten smokers gets lung cancer, therefore there must be a gene or genes; therefore we will develop a screening test and nine out of ten can smoke with impunity. That is obviously a false message. I do not know the extent to which the industry still is funding that kind of research. Certainly I would not accuse them of putting that out as a false message: I think they genuinely believed that this was a way out of the smoking controversy, as they called it. But there is certainly the potential. I think we have to remember when we talk about public health genetics that in that case there was a clear incentive to try to undermine public health by promoting a genetic approach to solving that problem.

**Q354 Lord Taverne:** This is a question, in the first case, for Mr Kent. You advocate more patient and family involvement in the evaluation of research proposals and their potential to deliver improvements. How would that work? How could this be done in practice? Are there examples of this kind of involvement?

**Mr Kent:** Yes, there are a number of examples that I can give you. For example, the Alzheimer's Society in this country funds a significant programme of basic and applied research. Historically they had the traditional expert panel of scientists and clinicians which reviewed applications for research grants that came forward to the society, but a number of years ago they set up in parallel with that a panel of patients and family members who also reviewed the research. They do not look at the technical adequacy of the research; that is the job of the scientific and clinical panel. Their job is to understand why the proposed research is important to families who live with Alzheimer's disease. As a result of requiring those people who seek funding from the Alzheimer's Society to explain to those people who live with the disease why it is what they want to do is important, how they want to go about doing it and what they think it will contribute to the progress towards the development of effective interventions and ultimately therapies, the clarity of the research proposals, the understanding

about the specificity of the objectives, just the general efficiency of the process, has increased significantly. At another level, orphan drugs, which I mentioned earlier, tend to be given a conditional marketing authorisation by the European Medicines Agency which requires further investigations to be done, monitoring their use in clinical practice. The Medicines Agency has invited representatives of the relevant patient organisation for the therapy which is being marketed to participate in the discussions with the company and with the representatives of the regulatory committee as to what is a reasonable degree of surveillance to generate the information that is necessary in order to establish continued evidence of safety and efficacy as opposed to that which might be desirable but which would impose too great a burden on the lives and circumstances of those people receiving the treatment. A number of other of our member groups, the Epidermolysis Bullosa Association, Cystic Fibrosis, the Muscular Dystrophy Campaign and so on, have patient and family representatives on their grant awarding committees, and that, again, tends to concentrate the minds of the researcher as to why what it is they want to do is important. How can they, as it were, justify the expenditure and the process that they adopt against the understanding of the families of what is important about the condition to them and to the situation in which they find themselves?

**Q355 Lord Taverne:** This does have an influence on the kind of research that is done, whether certain proposals should be adopted or not, and, also, presumably on the way in which research is done.

**Mr Kent:** Yes.

**Q356 Lord Taverne:** Which affects the researcher.

**Mr Kent:** Yes, it does. It helps organisations to identify clear priorities about what is the most appropriate way forward in terms of the needs of those affected by the condition, but, also, it helps to ensure that the protocols, the processes, the procedures that are developed

reflect and respect the needs and the situation of the patients participating so that they become more partners rather than just subjects.

*Dr Wallace:* There is an increasing amount of academic research on this idea of upstream engagement. It is being considered now also in terms of moving beyond just patients, because if you are looking at preventative work, preventing diet-related diseases or obesity and so on, then there is an argument that could be made to involve broader society much more; for example, asking people, “What would help you to live a healthier lifestyle?”

**Q357 Lord Taverne:** Since quite a lot of research has unexpected consequences and unexpected benefits, that is not inhibited by this.

*Dr Wallace:* I agree. You certainly would not want to omit all “blue skies” and just do what members of the public told you to, but you could certainly use it to inform your research priorities.

*Mr Kent:* On that very point, I think there was a suspicion amongst some members of the academic and scientific community that if you were to let patients have a view and express that opinion, the emphasis would shift to near-market or health services research away from basic research. In fact, it is quite the opposite. Because patients and families are interested in cures primarily, effective interventions, and in palliation or management of symptoms only if there is no possibility of a curative intervention, there is a strong commitment there to basic research as well as to the applied and the near-market stuff.

**Q358 Chairman:** From what you know, where is the research gap mostly?

*Dr Wallace:* There are a lot of research gaps in health, as I think you know.

**Q359 Chairman:** I was talking more about the social science side.

*Dr Wallace:* The second Wanless Report was very clear that there was a severe lack of evidence around public health intervention research, for example. There was a report commissioned which identified that something like four per cent of publications in the medical field addressed public health interventions. So there is a clear bias towards what can be patented, what is scientifically interesting, not necessarily what is going to deliver the biggest health differences. But I do not think I am the person to give you the answers to that; it is a question of finding processes that might give you the answer to that.

**Q360 Chairman:** It was the processes I was after.

*Mr Kent:* Again, I am not an expert in this field but we are very poor at understanding how to deliver health promotion messages. There is a lot that can be done, as it were, to reduce the risk or to mitigate the effects of complex diseases which have a genetic component. Some of that stuff, in a sense, we all know. It is common sense: it is the healthy living messages that are pushed out on a daily basis. But we do not understand, in a sense, how to communicate those messages to people/to families such that you can achieve a permanent change in their behaviour, so that even if they have a genetic predisposition to develop a complex condition they adjust perhaps the impact of lifestyle or environmental factors such as to reduce the likelihood of that risk turning into a progression into full-blown disease.

**Q361 Baroness Perry of Southwark:** Dr Wallace, in your evidence you submitted that you thought there should be legislation introduced to ban insurance discrimination based on an individual's genetic makeup. How different is that type of discrimination from current practices of insurers having access to individuals' family history?

*Dr Wallace:* I think there is a specific situation that arises with genetic testing, and I am thinking particularly of the predispositions to familial cancers – the BRCA1 and 2 genes (which the industry has always said is the test they want to look at next) and familial colon

cancers and so on. Research, for example, from Breakthrough Breast Cancer has shown that women do worry about the future insurance implications when they consider whether or not to take a test, so you have a specific circumstance where the medical decision that you take may be influenced by knowing whether or not the insurance industry will have access. I think that tends not to arise so much with things like cholesterol levels, where you know there is a known intervention that you are going to take. These decisions about whether to take the BRAC mutation tests, for example, are very difficult decisions. You are not sure how useful the information is going to be. You are not sure whether you are going to have a prophylactic double mastectomy (which is the main intervention you can take). When you are weighing up the difference between what the industry would say is the right to underwrite and what should be the individual's right to privacy in a medical situation, I think you can say very clearly that in those specific situations there is a real potential for insurance industry access affecting people's decisions. There is a real fear of that from the women involved. We know it is not going to impact significantly on the industry: under the moratorium they are not allowed to see those results and the insurance industry has not disappeared. There has also been research on that, doing the calculations as to how much money potentially they could lose. I think there is a very strong case there. Given that future tests will either be much less predictive or identify very small populations of patients, you could have something that parallels the current moratorium and protects the industry by saying, "Okay, if somebody applies for a very high value insurance policy then you can seek the results." I think that would work. There is a lot of evidence that it would work and that it would also reassure those individuals who are making those extremely difficult decisions about whether to take a test or not.

**Mr Kent:** I do not think there is a case for an absolute ban on this, because I think you would have difficulty in defining what you mean by a genetic test. In our view, the route by which you arrive at information is not important; it is the quality and the value of the information

that is revealed at the end of it. If there were to be a little fence drawn around the analysis of DNA, there would be a temptation to think that perhaps the industry then says, “Okay, fine, we have done that, we have tackled unfair use of genetic information in making the insurance decisions,” and carry on using other predictive forms of information in a much more relaxed way. In our view, any predictive medical information that is desired for use in insurance decisions should be evaluated very robustly, very rigorously, in order to establish whether the information was relevant, was significant. Is the person claiming to be competent to analyse the information and to draw inferences from it actually competent to do that analysis and to create the inferences? Is it significant in terms of the risk to the future financial well-being of the industry or the likelihood of a claim being made by the individual compared with everything else that is happening around? If you look at the attitude of the insurance industry to genetics, a few years ago they were very gung-ho: DNA testing was going to predict the precise moment of your death and the circumstances surrounding it in a huge range of different situations. They have ridden back from that. Although, as Helen says, there are applications in process in the Genetics and Insurance Committee for BRAC1 and BRAC2, they have been in process for a good many years now without coming forward with a dossier for the Genetics and Insurance Committee to look at. That would suggest to me that there is a considerable amount of rethinking going on in the industry about how valuable genetic information might prove to be. I should at this point declare an interest, in that I am a member of the ABI’s Genetics Committee, which does offer advice to the industry on the future progress of genetics. I would want that to be on the record.

**Dr Wallace:** I think what Alastair says about the industry is correct, that it is becoming aware that we are not all going to know the day of our death and that these tests are not going to be highly predictive. But I would make the opposite argument, that it allows an opportunity to legislate to protect people in this specific area of familial predispositions, and that there is still

a problem under the industry moratorium which Breakthrough Breast Cancer and the other cancer charities would tell you about: the issue of “test now, buy later”. There are women deciding whether to take the test now who do not know if they buy insurance later on in their lives whether at that point the moratorium will have ended and there will be a requirement from the industry. I think the industry at the moment wants to keep the door open just in case there is something it does not know about, and I do not think that is really a good basis to refuse to information.

**Q362 Chairman:** What is your comment about the recent United States legislation related to insurance?

*Dr Wallace:* If the US can take these kinds of steps then we should be able to take them in Britain. We would also argue that it should extend to employment to prevent genetic discrimination.

**Q363 Chairman:** To clarify, the United States legislation, GINA, is that the employer or health insurer cannot ask for genetic information but it does not forbid a life insurer to ask for information.

*Dr Wallace:* Yes, that is correct, but it does cover the employment situation, which we have not discussed. I think it would be good to move towards that happening in the UK.

**Q364 Chairman:** You are in favour of having such legislation in the UK.

*Dr Wallace:* Yes, to have legislation that would prevent both insurers and employers from genetic discrimination.

**Q365 Baroness Finlay of Llandaff:** Could I explore that a bit further. In the States, often your health insurance is linked to your employment, but we have an NHS here. I just wonder if you could separate those two scenarios out.

*Dr Wallace:* That is a very valid question. I think that is why we have not seen the same kinds of problems that they have already seen arising in the States, with people being refused coverage, for example, as a result of genetic tests. That is obviously a benefit of our system, but it does not necessarily prevent genetic tests being used more widely in the future. We know, for example, that the insurance industry does want to use the BRAC1 and 2 tests, and we know that genetic testing could become part of pre-employment screening for other reasons. That is something the unions are very concerned about that might be used to decide who gets a job in certain kinds of industries.

**Q366 Lord Broers:** Is there a danger that when dealing with common complex diseases genetic information may be seen by the public as more relevant in environmental issues and lifestyle choices?

*Mr Kent:* There is a slight danger but I think it is an addressable one. The fear that underpins your question, if I am interpreting it correctly, is that if you get a genetic risk identified then you will somehow develop the mindset, “I can do nothing about it, therefore there is no need to do anything about it,” and you will have this feeling that genetics is inexorably predictable and nothing can be done to alter your future health. With proper understanding and with proper information provision to people who are taking genetic tests, then the risk of that happening is very small. I think most people are capable of distinguishing between a certainty, a probability and an association, given the opportunity, and it is important that people providing genetic information do so in a suitable manner in order to make sure that people are not simply, for example, given relative risks but given absolute risks as well. Too often you see: “This result indicates that you have a 10 or 20 per cent increased risk of developing something.” Is that going from one in 100,000 to one in 90,000? The absolute risk is important as well, and we must clearly be careful to ensure that genetic information and information about other environmental and lifestyle risk factors is communicated

appropriately, so that people do not develop that kind of mouse-caught-in-the-headlights approach to genetic information but are able to evaluate it appropriately and effectively.

**Q367 Lord Winston:** Do you think there is a case that can be made for a bit better public engagement about this sort of issue? I am reminded of some news out today from the conference of the European Society of Human Reproduction and Embryology in Barcelona, press reports that in fact it is now going to be possible to screen human embryos for their genetic propensity for common diseases. In my view, a complete nonsense – but it is the sort of thing that gives alarm to some people because it starts to suggest that we can do far more which is determinist than we can.

**Mr Kent:** I would agree that there is a strong case for greater public understanding of the risks and benefits that are associated with increasing genetic knowledge but that needs to be put in the context, also, of our understanding of other determinants of health, so that we do not focus too exclusively on the genetic contribution to health and well-being at the expense of things which are much easier to adjust in our lifestyle and environment.

**Q368 Lord Winston:** If there is to be better public engagement, who should conduct it?

**Mr Kent:** There is a wide range of different routes or bodies which could do that. It can be done through schools, through the curriculum, with better science education and a better understanding of the role that science plays in our society. It can be done through bodies like the MRC and the Wellcome Trust, who have substantial public engagement programmes that, with more funding, could be rolled out to different communities. It could be done through patient groups, who have an important role in interpreting research information to those who are potentially sensitised to receive it, because they have a real need-to-know because they are affected or at risk by a significant health condition. If a way can be found to make it entertaining, then the media might even take it on – but we should not rely on the media to

communicate health messages, because, as I say, unless they are entertaining, they tend not to be picked up.

**Q369 Chairman:** There is this wide theatre doing exactly that just now, is there not?

**Mr Kent:** There is a lot of activity going on but it is not reaching significant sections of the public. Those people who are not in school are not reached by school-based programmes. People from minority communities tend not to receive standard health promotion messages. People who do not see that they have a direct need-to-know do not pick up information that may be around that may in fact be quite useful to them.

**Q370 Chairman:** Is this your perception or is this something that is evidence based?

**Mr Kent:** I think there is considerable evidence that public understanding of genetics is not as sharply focused as it might be, but they do pick up the sorts of ideas that Lord Winston has referred to and see this as being a more definitive statement of truth than in fact is the case.

**Q371 Baroness Finlay of Llandaff:** You just made me reflect that, until very recent times, public health messages in general, coming from people like the Chief Medical Officer, have categorised things as either safe or unsafe. Assessment of risk and understanding risk is a much more complex concept – a debate which perhaps Professor Calman instigated when he was Chief Medical Officer. I wonder for our inquiry how you feel that communicating concepts of risk and relative risk would best be played out to that large public that you just outlined. For school children, the reality of it is a long way off, and the need to know is a potent drive to learning.

**Mr Kent:** There is no one-size-fits-all model that you can adopt and say that if you just do this, then people will understand the idea of risk and proportionality and what-have-you. I think there have been a number of ways in which this has been tried. For example, in the

context of a genetic counselling clinic, counsellors will use examples, they will use analogies, they will convey things by using numbers or by metaphors. They will say, “This means about one person in a hundred may develop this, or you have a one per cent risk,” presenting the information in a variety of different ways so that it is possible for the person receiving it to picture it and to evaluate it and to incorporate it into their own understanding of their situation. Some people tend to like things given in numerical form; some people tend to like metaphors or analogies: “That’s about the number of people you can get on to a double-decker bus,” the sort of pictures that you can form in your mind to evaluate the issue and its salience to you. Really we need to try to make sure people are given access to information through a variety of media: written, verbal and in pictorial form, in order that they can take it in and assess it and process it, and have the opportunity to check back against an authoritative source if their understanding is correct.

**Q372 Lord Warner:** These are questions really for both of you. I am trying to wrestle with this issue of how you relay public understanding to the regulatory system. I want to go back a little bit to Mr Kent’s remarks about the NHRA being a little bit more energetic in this particular area. The public seem to me to be likely to have the understanding fostered if they can rely on the understanding of what the tests themselves fit. That is what they are up against: the issue of understanding, when the issue of taking a test comes up. If they are dealing with drugs, they have a guarantee that the regulator has not only tested the safety of the drugs but has tested the efficacy of the drugs as well, so it is easier for them, with a professional, to make a judgment. They do not have that same reliability, as I understand it, in relation to a genetic test because it is classified as a medical device. Do you feel that there is a fundamental flaw at the heart of all this in the way the tests are treated as a device rather than as a drug?

**Mr Kent:** Yes.

**Q373 Lord Warner:** Could you elaborate on what we can do about it?

**Mr Kent:** Referring back to what I was saying earlier, at the moment the In Vitro Diagnostic Directive simply looks at the technical adequacy: Does it do what it says on the tin accurately and reliably? There is no attempt to evaluate if what it says on the tin is important and useful. In our view, that second step should be part of the evaluation process. Because if you are somebody who is contemplating taking a test, knowing that the test is technically valid is important, of course it is; but knowing that the information it is going to reveal is useful and relates to the question you want answered is, in my mind, an essential part of the regulatory process. In the context of genetic tests, the important thing to know is the information that is to be revealed by the test: Can I trust it and will it tell me what I think I need to know about this particular situation? In our view, the NHRA should be encouraged to develop its input into the evaluation of tests, to look at that end of the equation as well as the technical end, which is currently all it is empowered to do under the IBD Directive.

**Q374 Lord Krebs:** Mr Kent, I would like to go back to the question about public understanding of genetic versus environmental risks. It seems to me that one phrase that is often used in media reports is the phrase “gene for”. “A gene for breast cancer has been identified” carries with it an implication of genetic determinism: “If I’ve got the gene I’m going to get the disease.” Do you think there is some work to be done around the vocabulary that is used. Just as Lady Finlay suggested we should avoid the word “safe” because nothing is absolutely safe, we should avoid the phrase “gene for”?

**Mr Kent:** I wish we could but I fear that train has left the station long since and I cannot see realistically how we can break that association in people’s minds. That is not to say we should not try, and perhaps there is a case for some research which looks at how messages are conveyed around the idea of risk and probability rather than causality in this area.

**Q375 Lord Broers:** Would there be a point in making a statement where one can approach being categorical? For example – and it is only anecdotal and my observation – a lot of people who are obese just relax back and say, “It’s in my genes”. That is endlessly said. Is that ever the case that it is in their genes or is this just a convenience? Should a statement be made about that – that this is just not true or very, very rarely the case?

**Dr Wallace:** We published a report in 2006 on nutrigenomics, the idea of tailoring your diet to your genes. One of the things that came out of that is a very clear indication, for example, that all the genes that had been linked with obesity either are very rare or have not been replicated, or, in the case of a few recent ones, they are involved in appetite. It may mean, therefore, that an individual may eat slightly more than another, but it certainly does not mean that some people can eat what they like and not put on weight – that ongoing myth about metabolism. Earlier research in the metabolism area has been refuted, but there is no scientist or anybody else whose particular interest it is to be the person who goes out there and explains to the public that that research has been refuted or that this myth is no longer widely believed in the scientific community. I think there is definitely an area for messages around that. I would agree strongly with Lord Warner’s point that one of the issues around the marketing of these tests that we see, many of which are being marketed with supplements and diet-related advice, is that you have got this very strong message in advertising now and on the Web that may be giving people misleading messages and unless you address that and address the regulation issue then anything you try and do on the public education side is likely to get swamped by these alternative messages. I think it is very harmful particularly in terms of potentially confusing people. *Which?* did an investigation last week on ancestry tests where they found that three different companies gave three different genetic ancestries based on the same sample. We know this is happening with the diet-related tests. People have not really woken up to that yet but if you get a situation where you are getting widespread tests

and some people with the same sample are being given completely conflicting results, information and advice then there is a real potential of making the difficulty in getting public health messages across that we have now much worse.

**Q376 Chairman:** Thank you very much. We slightly overran our time but before we finish what two recommendations are you expecting out of our report?

**Dr Wallace:** I would like to see a recommendation for statutory regulation of the clinical validity and utility of tests so that both those aspects are assessed before the tests can be marketed.

**Mr Kent:** I would like to see a recommendation coming from this Committee that in looking at the future of genomic medicine and its potential to change things for the better for families living with intractable disease, proper consideration is given as well as to the risks and benefits associated with doing the research to the risks and benefits that may follow from not doing the research, because it is not as if we live in a situation where everything is for the best in the best of all possible worlds now. People are living with intractable medical conditions. This research may deliver answers for some of those conditions and we need to make sure that that is fostered and there is a cost that is associated with not doing it that needs to be established and borne in mind.

**Q377 Chairman:** Thank you both very much for making time to give us this evidence, it is very helpful. Thank you.

**Mr Kent:** Thank you for the opportunity.

**Memoranda submitted by UK Genetic Testing Network  
and London Specialised Commissioning Group, Bexley Care Trust**

Witnesses: **Professor Peter Farndon**, Consultant Clinical Geneticist and Director of the UK Genetic Testing Network; and **Mrs Jacquie Westwood**, Director of Specialised Services for South East London, Bexley Primary Care Trust, examined.

**Q378 Chairman:** Good morning to both of you and thank you for coming to give evidence, Professor Farndon and Mrs Jacquie Westwood. Again, if you have any opening statements, please feel free to make them. Would you introduce yourselves as to who you represent when you first speak. Can I invite you if you have any statement to make to make it.

**Mrs Westwood:** Thank you for inviting me. My name is Jacquie Westwood, as you have heard, and I am representing commissioning in the NHS. I work for the London Specialised Commissioning Group based in Bexley Care Trust and part of my role, which has been agreed across the ten specialist commissioning groups in England, is for me to work with the UKGTN as a project director in collaboration with all stakeholders in order to secure the best possible service and equity for NHS patients.

**Professor Farndon:** I am Peter Farndon. My day job is Consultant Clinical Geneticist in Birmingham so I actually see patients, but I am here today as Chairman of the United Kingdom Genetic Testing Network for which my foundation trust allows me time to come out and chair the meetings. I think UKGTN has been extremely successful in delivering for patient care tests that have been identified both by patients and by laboratories and clinicians. I think it has been so successful because it has been grounded in NHS practice so that we have actually managed to identify what needed to happen and then got somebody to pay for it in the NHS. I wonder if you would also allow me to explain what we mean by genetic testing. In all the work of the UKGTN genetic testing means two separate things rolled together in that term. The first is the laboratory assay which has to be quality controlled and delivered -

by whom we can come to later - and then there has to be clinical interpretation of those results and an explanation put into a clinical context. The UKGTN definition of genetic testing is both those two bits of explanation and the laboratory assay - and we think that is really important in thinking about this. The results of this process mean that some of it has value for individuals and some of it has value for families when you set it in a clinical context. The other thing that we have come to realise is that when we started work the results of our genetic tests had absolute values to patients - yes/no answers - because they were for single gene disorders. In the UKGTN in discussions about the future and use of SNPs and predictive tests and things like that, we have come to appreciate that it is all about relative risks really and how you put the relative risks from a genetic test against all the other risks that the patient comes into in medicine and their disease, so their age, sex, what they eat, where they live, and their population ancestry. We have got two ways of thinking about genetic testing. One is absolute risk and one is relative risk but the most important thing is for both of those two things what does it trigger in health interventions or information.

**Chairman:** We might come back to that but can I ask Lord Winston to start.

**Q379 Lord Winston:** Genetic tests are now available for which it is claimed you can predict common diseases, you can predict drug efficacy and toxicity and guide therapy in diseases such as cancer and leukaemia. What do you think are the best ways to assess the clinical validity and utility of these tests?

**Professor Farndon:** If we go back to the UKGTN model which we have used for single gene disorders, we think that the model is still valid.

**Q380 Lord Winston:** Can we exclude single gene disorders.

**Professor Farndon:** Yes, but we think that as part of that process there has to be the scientific validity of the test and therefore the scientists and epidemiologists at the moment

are coming up with gene disease correlations, so we are having the scientific data but then what has to happen is the clinical performance, and for the NHS that is really important because that has to be paid for. We think those two things have to be taken into whatever system is used to assess that test. You need a system to identify the scientific validity. That can come from studies with the MRC or the Wellcome or various other things but then there needs to be a bit which is not present at the moment to determine how that applies in clinical practice. That is the gap at the moment.

**Q381 Lord Winston:** Which of these new tests do you think will have the most impact on the NHS first?

*Mrs Westwood:* Can I say both, because I think the clinical validity is important because it is assuring the quality of the test ---

**Q382 Lord Winston:** I am sorry, I did not ask my question quite clearly enough. Which new genetic tests will have the most impact on the NHS?

*Professor Farndon:* The information that comes from a combination of SNPs probably in areas like the combination (and I cannot remember how many it is, eight or so) SNPs for the variants in breast cancer and the ones that are associated with increased predisposition to prostate cancer for instance, I guess what will happen is when they have been sorted out the combination of the eight SNPs in the low penetrant genes for breast cancer will move a woman's risk up and down the screening programme at the moment. A certain combination will say, yes, you need screening at the age of 50; another combination will say, together with all the other lifestyle things as well, you do not need screening until you are 70 because of the relative risk. UKGTN thinks those are the kinds of combinations and maybe there will be combination for hypertension or a combination for diabetes but of course we do not know at the moment.

**Q383 Lord Winston:** People like James Scott have spent most of their recent research life trying to investigate the genetics of diabetes and have drawn a total blank.

**Professor Farndon:** I was a great sceptic over this and I have been for a number of years, as some people will know, but there are certain times when one goes along to a scientific conference and you can see that things have actually moved, and at the last European Society for Human Genetics, combinations of SNPs and genes were suddenly being presented from the Wellcome Case Control Consortium and some of the work that is coming out of Hinxton as well looking at the cancer genes which make me more who hopeful. However, I cannot give you a timescale. The most important thing is that we have a mechanism in place to decide how we will deal with this when this information is available.

**Q384 Lord Winston:** And which body do you think should cover these new types of test, the UK Genetic Testing Network?

**Professor Farndon:** I think it should be a body that can horizon-scan, that can see what is coming along, that can make sure that scientific validity tests have been performed and then can undergo a clinical assessment about performance. UKGTN does that at the moment for single gene disorders. What has also become apparent is that the borderline between cytogenetics and molecular genetics is very blurred at the moment, it is a matter of scale, so we have started looking at arrays and how do we get those into clinical practice. The UKGTN would have something to offer, the process that we have developed would have something to offer, but I think it is a wider debate about what such an organisation would look like.

**Q385 Lord Winston:** I want to be absolutely clear and I want to press you on this because this Committee will in due course be advising ministers and government about what action it should take in an extremely complex and very, very expensive area. Are you really confident

that the analysis of SNPs is really going to make a fundamental difference to healthcare in 10 or 15 or 20 years' time?

**Professor Farndon:** I am more confident - it might not be analysis of SNPs, it might be copy number, it might be sequencing - that there will be more information coming out of the human genome in very particular cases.

**Q386 Lord Winston:** Without any evidence about gene expression for example?

**Professor Farndon:** One hopes that that will be the way it will go, that will supersede all this and you will find out how the genes actually work.

**Q387 Lord Winston:** That would be very complex and expensive.

**Professor Farndon:** Yes, which is why it is even more important that the clinical performance of these tests is sorted out before we decide whether or not to go down there. It might be that such an organisation says on the basis of all the information, this is not something we should be doing, but the problem at the moment is that we have not got a mechanism to decide that.

**Q388 Chairman:** You did not say, in response to Lord Winston's question, which organisation you think should be the one that takes it on. You partially answered that question that you were doing it now for single gene disorders but have you got the capacity to do it if there was a need - and we are not quite sure that there is a need - for more common diseases, and what about the role of NICE and MRHA?

**Professor Farndon:** We have discussed these in UKGTN about the different organisations. Jacquie, do you want to say something?

**Mrs Westwood:** We understand that there are a number of organisations that are appraising whether services should be of value to the NHS and that is very important from

commissioning because it is giving you an assurance about what you commission. I think for the area of pathology in the widest sense there has not been the same focus and perhaps this is because the NHS in general does not commission for pathology in the same way as it commissions for other services because pathology has perhaps traditionally been seen as an infrastructure within a hospital so that cost is associated in different ways. I think in terms of evaluation, what we have found in genetics where commissioners have had a real role in commissioning tests, is that it has been extremely important for us to find a process by which we have assurance of the clinical validity and utility as well as the scientific requirements and by designing the process we have that, so in terms of pathology in the widest sense, my view is, as Peter has explained, the process that we have developed is transferable. It does not necessarily have to be the UKGTN that would take that on board but there are many areas where the boundaries merge and cross and therefore, whatever happens, the UKGTN in its role would have to be able to collaborate with partners in dealing with that, so my view is with the Department of Health recently setting out its Diagnostic Programme Board, we should somehow be able to use that in order to design a process which fits them all together because, if not, I think decisions will not be made in a consistent manner. That is absolutely important because otherwise we will get into lots of duplication which is unhelpful, so my view is that UKGTN could have a role in helping this in terms of the process. Clearly there would have to be additional expertise available in order to make those processes work.

**Q389 Chairman:** Let me move on to question that follows. If we have done all the research and we find that a test has utility and has been found to be cost-effective and now needs to be introduced in clinical practice, how should that be done so that it does not end up that it is introduced in some areas and not in other areas and who should do that?

**Mrs Westwood:** Again we have been able to achieve that because, as Peter has said, originally ---

**Q390 Chairman:** But that is only in London.

*Mrs Westwood:* No, it is national, it is UK, England, Wales, Scotland and Northern Ireland.

**Q391 Chairman:** Which tests, can you give us an example?

*Mrs Westwood:* Any test. The new genetic tests which have been evaluated have been introduced nationwide.

*Professor Farndon:* About 350 of them I think.

**Q392 Chairman:** There are currently 350 genetic tests being used in the NHS?

*Professor Farndon:* No, these are the ones that have been assessed and are paid for by the commissioners.

*Mrs Westwood:* How we are able to do that is by making the whole thing connect so that, as Peter said, it is embedded in the NHS. It is not something on the side, it is real business, and therefore it is very important from a patient perspective that we achieve this equity, so what we have been doing is ensuring that through the processes of engaging with commissioners as well as with our clinical and scientific colleagues we have got design processes by which these are looked at every year and agreed or not agreed in order to be available in the NHS.

**Q393 Chairman:** What is the uptake like?

*Mrs Westwood:* In terms of the uptake of tests, individual commissioners are reviewing that and they feed back to the Department of Health group which has been set up in order to see. We are looking at that again from the UKGTN perspective to actually look at the uptake. I do not have the figures as yet but we are looking at the uptake. What we have found is that the predictions that had been made, both clinically and from the scientists, are normally fewer than their original predictions that are provided at the end of the day.

**Professor Farndon:** There are some policy tensions here because what as Chairman I am desperately keen to make sure is that we have equity of access to the existing genetic tests that are part of this network. We set that up by agreement and consensus nationally and then have to rely on genetic laboratories in foundation trusts to provide the service, paid for by commissioners in specialist service groups and PCTs, so there are some policy tensions within the system.

**Q394 Lord Warner:** Can I just explore some of these policy tensions, as you slightly euphemistically describe them. Can we just be very clear about how this commissioning system works. You identify - this is a query - 350 tests at the beginning of a year and you ensure their availability through reputable providers of those tests, and you do that on some forecast basis by the NHS, and then you rely on people in the NHS to use these commissioned facilities on a day-to-day basis? If Dr Winston needed one of these tests he would go through that commissioned 350 tests? Is that how this works? I think it is quite important that we understand how this works.

**Professor Farndon:** There are 352 diseases that are paid for by commissioners under the specialist services definition number 20. There are of course other laboratories which provide other tests which give genetic information and it may be that there are different systems for paying for those. The diseases and tests that are provided through the United Kingdom Genetic Testing Network are in laboratories that have come to a certain standard, agreed that they will be involved with the clinician and have agreed what information the patient will get at the end of it. They are paid for through the specialist commissioning service.

**Q395 Lord Warner:** If you are in the NHS for one of those 350 disease conditions, you cannot get your test done other than through had that network? Or are those the policy tensions you are talking about?

**Mrs Westwood:** From a commissioning perspective, the members of the UKGTN, that is the regional laboratories and services that are part of it, come with a quality assurance, so we know from the patient perspective ---

**Q396 Lord Warner:** Sorry, I am not making my question clear. I am talking about then we get back to the individual doctors dotted around the NHS. Does this system mean that they can only get tests done for those 350 conditions by going through one of the approved laboratories that you have identified?

**Professor Farndon:** Unless they wish to say, "I will go to a laboratory overseas," or, "I will go to a non-accredited laboratory in this country if somebody else provides it," and that is their decision because they are paying for the test. If they want it to be paid for through the specialist commissioning service then they have to go through one of these laboratories. The tension we have got is if an ophthalmologist wants to send a test in, they have no funding stream in ophthalmology to pay for it unless they pay for it out of their budget. The funding stream for the majority of these tests is through the genetics services; that is another policy tension. If we try to roll out equity of genetic testing into other specialties, we have to come to some re-think about how that might occur.

**Baroness Finlay of Llandaff:** Before I ask you my main question I would just like to follow up on that very briefly over these policy tensions that you refer to because we also have devolved health administrations and I wanted to ask you how your principle of equity works across the devolved health administrations as well.

**Chairman:** Where in some of them there is no commissioning?

**Q397 Baroness Finlay of Llandaff:** Yes, exactly.

**Mrs Westwood:** Except that they are part of our group and they send members to the steering group. Our scientific adviser is based in Scotland. The devolved countries use the advice that

we give to commission services nationally for their services as well, so we do have some genuine consistency.

**Q398 Baroness Finlay of Llandaff:** Are you buying into Wales and Scotland as well as Wales and Scotland buying out?

*Mrs Westwood:* Yes, absolutely.

**Q399 Baroness Finlay of Llandaff:** Could I go on to the main question I wanted to ask you which is really how the NHS should plan for the new types of genetic tests coming into clinical practice and as these tests emerge how the requirement for the test can be measured and anticipated.

*Professor Farndon:* At the moment there is no system of planning because it is about horizon scanning and seeing what is going to come up. At the moment it is generally technology-driven, so a new type of genetic test or a new assay comes along, a group of people in the NHS take that on, work it up, convince clinicians that it is worthwhile and if it is a genetic test they will, with the information, present it to the UKGTN for the single gene disorders. In the future we cannot see a mechanism for, say, a combination of SNPs and where that would fit, whether each individual cardiologist would be targeted by a laboratory who was providing this to say “buy it from us” or who would say you actually need this test. It is clear that if there were a commercial provider that commercial provider would offer it to the NHS and then decisions would need to be taken about whether the NHS bought it or not. We cannot identify a mechanism within the NHS structures that would be able to commission this on a national level.

**Q400 Baroness Finlay of Llandaff:** So where should that sit looking into the future?

*Mrs Westwood:* Again, I think it goes back to the question that was asked previously about what sort of body you have in order to oversee some of these things. I think they are linked in terms of the questions and, as we said, we can play a role in that, but the most important thing is having the process in order to do that evaluation once these things emerge because we cannot predict everything so what we need to have is a process which deals with them as they emerge which is rigorous, and I think that is where the benefits come rather than thinking all the time about what is to emerge.

**Q401 Baroness Finlay of Llandaff:** How do you think we should be planning though for the national and local tariffs for such tests and the costs of them in relation to the money that they will then take out of existing clinical services?

*Mrs Westwood:* I think that is one of the policy tensions that is around because at the moment there is no proper understanding of the way that genetic services are commissioned nationally. They are all dealt with differently in the different areas and there is no structure to that and therefore the tariffs do not apply because everybody is doing it differently, and so therefore in any reference cost that you collect it is always, I am afraid, apples with pears, and it is not consistent. This is one of the areas we have been trying to work very closely with the Department of Health in order to look at how tariffs can be constructed to see that we also have value for these new tests as they come out and to know whether they are aligned with the specialty, which they are in some cases, or whether they are aligned with genetics which they can be in other cases. Bearing in mind that of tests ordered, some 70 to 80 per cent are ordered directly from physicians rather than through clinical genetics, it is a very important issue to sort out.

**Q402 Baroness Finlay of Llandaff:** I would like to follow up on Lord Winston's question to you earlier thinking about the health benefits of these tests because I wonder whether we

are putting undue emphasis on genetics versus environmental and infective agents in disease and disease development, particularly because the latter are potentially much more modifiable and would be affecting gene expression whereas looking for the substrate gene is something that we cannot affect and we still do not know what alters expression. I wondered what your comment is on that. I recognise that you have a vested interest one way.

**Professor Farndon:** We only have a vested interest in making sure that families with single gene disorders get the services and will continue to get the services. With regards to the future about how we look at genomic information, I hope we have given the impression that we have quite an open mind actually. The only way that you are going to find out whether this works is to do it and model it and find out.

**Q403 Chairman:** I think it is an important point though, leaving aside, as Lord Winston says, single gene diseases when we talk about common disease association with SNPs variations and it pushes up the risk overall for an individual that has, let us say, five or six SNIP variations from a general risk of, let us say, 1.2 per cent to 1.25 per cent, what does that mean?

**Professor Farndon:** Absolutely.

**Q404 Lord Warner:** Could I move us on to pathology laboratories. The use of genetic tests is increasing, as we all know, and that is increasing both in the regional genetic centres and in NHS Pathology Service laboratories that have not traditionally specialised in genetic testing. We have heard in evidence that future pathology services might usefully be re-organised with a molecular biology lab acting as the hub of molecular testing, clinical genetics, cytology, haematology and pathology. What do you think would be the benefits of organising pathology services in this way and do you have any idea what the cost would be and what would be the desired timescale?

**Mrs Westwood:** I think there is a bit of a dichotomy because in terms of the actual testing that is done, it is the molecular technique that is used in pathology in the main rather than a genetic test and therefore it is the technical platform which is important in terms of its generic usage. I am absolutely clear that there are efficiencies to be gained by organising things differently. As a commissioner I am faced with this in terms of how the trusts themselves want to organise themselves. What I have found is that I have to ensure that the clinical scientists' input into the clinical requirements of genetics is maintained so that you can have an approach where in terms of pathology the factory can do the high throughput test with much more efficiency, however, because in genetics currently the volume of tests is much smaller - extremely small, it is not high volume and it requires an interpretation very clearly but with the scientists working with the clinicians - therefore in terms of its organisation where I have had to face these issues I have been working with the trust because there are very few regional genetic centres to make absolutely sure that the clinical scientists continue to work alongside the clinicians.

**Q405 Lord Warner:** As the number of these tests that become available in the NHS increases does it become inevitable that you move to a commissioned service rather than treating pathology, as you rather elegantly put it before, as basically an infrastructure overhead?

**Mrs Westwood:** With pathology the only thing PCTs have commissioned traditionally is the direct access tests from their GPs which are obviously the very basic tests that are undertaken through pathology in general terms, so with genetics it is very different and also with screening it can be different. If I commission blood spot programmes for London, because we have three regional laboratories that are doing those, from my perspective I think it is really important that we still gain these efficiencies by critical mass but look very carefully at its organisation so that we do not lose the clinical advantages we have currently.

**Q406 Lord Warner:** I do not know whether Professor Farndon wants to come in on that.

**Professor Farndon:** I think we all agree that we need high throughput sequencing facility for the UK. The White Paper went some way to making sure that we had some capacity in the existing genetic services, but if we move towards SNPs and pharmacogenetics we are going to have a high throughput sequencing somewhere and that is what, as far as I can understand it, these laboratories will do. They will give you the power to put lots of specimens through. If you had an algorithm to work out from the results you got the answer for whether you prescribe this drug or that drug or this is the relative risk, that would be fine, but if we were to do this we really need to make sure that there is still a process in place for picking up how you work out unusual variants in genes of high penetrants. The way that is done at the moment, with excellent clinical results, is making sure that the clinical scientists work very closely with the clinician who understands the disease and usually with a research group as well. If we were to go to this process and put high throughput sequencing for single gene disorders in such a unit, we really must make sure that the research and clinical links are established otherwise we will degrade the service for patients.

**Q407 Lord Warner:** Does this mean that a lot more new equipment is going to have to be put in place in particular areas, or is the new kit that has been put in likely to be adequate?

**Mrs Westwood:** The new kit that is currently going in has not as yet been proven in terms of the higher throughput sequencing. For the kit we are using at the moment from the White Paper money (and considerable investment was put into kit in the regional centres) it took at least two years in order to get the value out of that equipment and it took longer to install and commission than people thought. In order to prove that the results that you had were clinically accurate it took much longer than people thought.

**Professor Farndon:** And for some genetic disorders it is not producing the sequence that is the hold-up. For instance, we are trying to deal with the backlog of BRCA1 testing for breast

cancer predisposition, and it has been the interpretation of the results and whether this variant actually means that it is a pathogenic variant, and you have to look at what that does to the protein and sometimes express it in vitro and other times you may need to follow that through members of the family to decide. Putting it in a large laboratory will give us some economies of scale but we still need to make sure that for certain genetic disorders these other skills are available.

**Q408 Chairman:** I thought Lord Warner's question was about focusing on laboratory services as opposed to who orders the genes, so it is the laboratory side that we are trying to get a grip with, whether that would be better done by a centralised hub.

**Mrs Westwood:** I think we were saying that you could have a factory environment where you are doing the laboratory element of it but because of the interpretation of those results, the clinical scientists at the moment have a dual role so it is possible to have more of an environment for the high throughput areas in that environment but we still need to assure the interpretation because the software is not developed to be intelligent; it requires the person to interpret the results at this moment in time.

**Professor Farndon:** So you would have your high throughput sequencing machines but you would then need a group of clinical scientists maybe working in that unit to take the results of some of those and do much more detailed work on it. If you put them away from the clinical side and the research side we fear that that may degrade the eventual service to the patient.

**Q409 Lord Warner:** Are you saying that if you want to get the economies of scale for the kit, if I put it as crudely as this, you have got to have the interpretive clinicians near the kit?

**Professor Farndon:** You have got to have interpretive clinicians with a group of other scientists who can take the information and interpret it and maybe do some more laboratory experiments and techniques on it. You either put all those together in one place or you put the

kit in one place and then this other little laboratory somewhere else which is against the spirit of what you are trying to do.

**Q410 Lord Warner:** I am not trying to do anything. I want your professional views on which makes the most sense clinically and economically; you are commissioning this stuff?

**Mrs Westwood:** I am.

**Q411 Lord Warner:** You have a lot of experience and we are trying to get a feel for what you, based on your experience, would see the future as being with a big surge in the number of genetic tests

**Mrs Westwood:** And in working with services in London, in terms of them reviewing how the pathology services may be provided in a single organisation, not necessary across a whole network but where perhaps they might have over 20 laboratories in a particular organisation, what I have agreed with them in order to protect and to ensure equality of service for patients is that the scientists will have a dual role, one to work in setting up the technical aspects of the test but then they will continue to work with clinicians and the academics in the interpretation of those results to assure the quality of the service for clinicians.

**Lord Warner:** Thank you very much.

**Q412 Earl of Northesk:** We have heard that updating of IT systems is required for efficient analysis and interpretation of genetic tests, and for transfer and storage of genomic data. As testing and use of genetic information within the NHS increases substantially, what features should be built into these IT systems to make full use of the information available from genetic testing?

**Mrs Westwood:** We have a fundamental problem with genetics because it is not classified in terms of the NHS and therefore if you look at the NHS system for coding disease, we cannot

effectively and have not been able to have an information base about genetics, and that is a major issue, particularly if we want to have valuable information around population in a general sense, and therefore it requires a lot of work. We have been working very hard in the UKGTN on this network to work with colleagues both in Europe and in America in order to get some classification system. At the moment we have got very limited OPCS, which is the lowest level coding, and nothing really in terms of HRG, so we really do not know what is going on. Of course the diagnostic element is not coded either. What makes it more complex for genetics is that the NHS deals with individuals and not with families and so that is another issue to be addressed in the widest sense. We have been working very hard in numerous organisations since the NHS has changed its organisational status around IT to try and resolve that issue and we continue to do so. I have to say that what we have done or been able to do in conjunction with one of the reference laboratories which is Manchester, which took the lead, was to be very helpful in determining the data requirements to enable these changes to take place. We have continued to want to work to agree the data requirements in order to fit within the NHS framework, but there is a long way to go.

**Q413 Earl of Northesk:** How high up your list of priorities is this particular issue in the scheme of things?

*Mrs Westwood:* Yes.

**Q414 Earl of Northesk:** Very high?

*Mrs Westwood:* Yes. We summarised in our return the key issues that we felt and that was one of the issues that we have summarised in terms of needing attention. Again, we are happy to take that, remembering that the UKGTN does not have an organisational status, it is purely a network and purely is influencing and negotiating in a way with no real organisational responsibility so I think it is seen as helpful because in a way it is independent.

**Q415 Baroness Perry of Southwark:** We have been cautioned against the view that genetic tests are seen as different from tests for other biomarkers - I think it is known as 'genetic exceptionalism'. How should the provision of information to clinicians, policymakers and the public be tailored to guard against that view?

*Professor Farndon:* We were fortunate in hearing some of the things that Alastair Kent said earlier on and we agree there is a great danger in genetic determinism and we do not subscribe to that. You will notice that when I explained what we thought a genetic test was we said that it was between the analysis and the information that is given and I think the key to all of this is explaining what you expect the test to give you. Sometimes it might say that you should eat less broccoli and other times it will say "You are going to get Huntington's Chorea; I don't know when but you will if you live long enough." I think in talking to patients in the clinic, they can see the difference actually, and there is a feeling that the NHS is good for important medical things and people will talk to the NHS when it is important and maybe treat information about broccoli and how you detoxify alcohol in a different way. That is just a feeling we get through asking patients in a clinic about how they see these different kinds of things. I think this is all about the information that the providers of tests give to people. I am not sure that legislation about over-the-counter testing or legislation about genetic testing would solve the problem because the Internet is out there and people can just buy results from all over the world. What I think is a three-step process, first of all, before people send their sample off they need to have consented and realised to what they are consenting; the second is it must be clear that there is a quality control laboratory; and, thirdly, an explanation of what this information actually means. This goes again to relative and absolute risks.

**Q416 Baroness Perry of Southwark:** So you would go for the California ban really?

*Professor Farndon:* No, I do not think I would go for an outright ban.

**Q417 Baroness Perry of Southwark:** It is not an outright ban, it is based on those conditions that you have just described, that it has to come from a registered, accredited laboratory and there has to be interpretation by a qualified medical practitioner.

**Professor Farndon:** I share some of Alastair's concerns that if you ban it outright --- should you ban people finding out if they are going to react to broccoli? Some people argue and some of the patients have argued with us at UKGTN that it is their right to know that information and "why are you so paternalistic as to not let us?" so I think we have taken the view that the laboratory assay is the same in looking at the sequence of DNA but what you do with the information is different, so therefore the single gene disorders are regulated under the NHS and who pays for them. I would have thought a code of conduct would suffice but the most important thing is the effect that the Advertising Standards Agency could have.

**Chairman:** I thought the Californian law is that you can have your gene identified and sequenced but the interpretation of it, what advice that you should follow should come from a medically qualified person. That is what the Californian law says.

**Baroness Perry of Southwark:** And an accredited lab.

**Q418 Chairman:** You do not agree with that?

**Professor Farndon:** I am not sure about that.

**Q419 Lord Colwyn:** I think your evidence this morning and certainly your very helpful papers have stressed the uniqueness of the UKGTN network and the gene dossiers for the new genetic test being proposed for NHS services. Are there differences between recommendations for genetic testing carried out in NHS laboratories and in the private sector which I think earlier Alastair Kent said is in a state of confusion at the moment. Could you say how these should be best managed.

**Mrs Westwood:** We have a facility within the UKGTN for any technical provider, ie not with the full clinical service, to provide the test so they can apply to the UKGTN, and indeed we have had a technical provider, so if they wish to work with us they can, and normally they are working with a genetic service to provide that test because of the particular expertise or skills that they have or equipment that they might have in order to provide it. We certainly do not exclude other providers supporting the work that we do, but we do ask them to have standards so that we are assured of the quality. We do not ask them to have a standard of quality but it has to be a recognised quality agreement and that has not been a problem for the provider that wanted to apply.

**Q420 Lord Colwyn:** Standards are monitored, are they?

**Mrs Westwood:** Yes.

**Professor Farndon:** As far as UKGTN was concerned, we took the view that it did not matter who provided the assay as long as it was scientifically valid.

**Chairman:** If members do not have any more questions, thank you very much both of you for your contributions today; it has been very helpful. Can I say that if on reflection you feel you could supply some more information related to the questions today, feel free to write to us; we would welcome that. Thank you very much indeed.