

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

WEDNESDAY 7 MAY 2008

PROFESSOR SALLY DAVIES, DR MARK BALE, MS DIANE PAYNE,
MR PAUL WILLIAMS and DR SIVASEGARAM MANIMAARAN

Evidence heard in Public

Questions 59 - 121

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WEDNESDAY 7 MAY 2008

Present

Colwyn, L
O'Neill of Bengarve, L
Patel, L (Chairman)
Perry of Southwark, B
Taverne, L
Warner, L
Winston, L

Witnesses: **Professor Sally Davies**, Director-General, Research and Development and Chief Scientific Adviser, **Dr Mark Bale**, Deputy Director of Scientific Development and Bioethics, and **Ms Diane Payne**, Team Leader, NHS Genetics Team, Department of Health, **Mr Paul Williams**, Head of DIUS Research Council Unit, and **Dr Sivasegaram Manimaaran**, Innovation Policy Directorate, DIUS, examined.

Q59 Chairman: Good morning. Thank you for coming to give evidence today. May I welcome the members of the public. You will find information, including a declaration of interests of the members and who the members are. I presume, Professor Davies, you will perform the lead role. Before we start, could I ask each of you to introduce yourself for the record and say what your position is and which organisation you represent.

Professor Davies: I am Professor Sally Davies. I am the Director-General of Research for the Department of Health and have responsibility for the National Institute of Health Research as well.

Dr Bale: I am Mark Bale. I am Deputy Director of Scientific Development and Bioethics in the Department of Health with responsibility for genetics, stem cells, and matters of ethics.

Ms Payne: I am Diana Payne. I am Team Leader in the NHS Genetics Team, Department of Health, chiefly responsible for the policy on NHS genetic services and for implementing those elements of the genetics White Paper as well.

Mr Williams: I am Paul Williams. I am head of the Research Council Unit in DIUS, where I am responsible for the seven Research Councils.

Dr Manimaaran: I am Sivasegaram Manimaaran. I am a team leader working for the Technology Strategy Board working inside the Innovation Policy Directorate.

Q60 Chairman: Thank you very much. I see that you have a strong team behind you, Professor Davies, to support you. You are all welcome. Each of us will have a question or two to address to you and you can decide who should answer. Before we start, do you have any statement you would like to make?

Professor Davies: I would like to say on behalf of the departments that we do welcome this inquiry into genomic medicine and we are looking forward to your report – and on this occasion I mean it – because it could be very helpful in determining the future direction of the Government’s initiatives. As you know the White Paper in 2003 *Our inheritance, our future* set out the Government’s vision for genetics in health care and the recent progress review was sent to you. That highlights the challenges that we face in the future, as well as many of the achievements, such as the setting up of the NHS Genetics Education and Development Centre and, even, a week ago, the announcement of a successful gene therapy for blindness and other gene therapy trials. The education and development centre is working to get appropriate levels of genetics knowledge into the education and training of the NHS workforce, so there is a lot of work ongoing that your report has an opportunity to influence.

Q61 Chairman: Thank you very much. Does anybody else have any comments? Okay. From the evidence that we have had – and it is quite voluminous evidence – and also evidence

that was presented to us at a learning seminar that we had, it is quite clear that genomics and genetics and therefore the implications for health care and to medicine, are moving along at a pace and that the potential for genetics in health care, therefore, in the coming five years or maybe, at the most, ten years is going to be quite significant. First, would you agree with that assessment? Second, if you do, what do you see are the key challenges, particularly in terms of health care reorganisation that will be necessary to deliver genomic medicine. Are the Government anticipating these developments in genetics? How is the Department preparing for this? What do you think are the changes that will be required?

Professor Davies: Thank you, My Lord Chairman. We would agree with you that the potential is absolutely enormous. How will genetics impact on health care? Through finding genes or their expressions; to give profiling for people, either as patients or even as a population, for screening that will lead to prevention of disease/health promotion. Genetics has already started to have a role in diagnostics. In disease management there are genetic tests which direct which way we should treat people. There are early pharmacogenomic tests to look at the safety and efficacy of drugs, and of course there are treatments that are based on genetics. The one that I know most about is chronic myeloid leukaemia and how genetics contributed to making the oral drug that links to the protein kinase receptor, let alone the gene therapies. Genetics in health care will need to play into every area of what we do. We feel that the White Paper was a good start at that, highlighting initiatives to develop specialist capacity; but it is clear that we need not only clinical genetic specialists but that genetics will become the province of all clinical professionals over time as it moves out from the specialist area. You only have to look at the Wellcome Trust case-controlled consortium data to realise that no part of medicine will remain untouched. We ourselves are contributing with the MRC and the Wellcome to the UK biobank study, UK Biobank, and clearly that will engender more data that we will need to use. What are the challenges? In summary bioinformatics is the first

challenge, both for research and the science underpinning it but also as we want to bring genetics into the health care arena. We are well supported in this country, having the European Institute as well. There will be the issues of embedding genetic technology and knowledge into the service and into clinical practice and that takes one back to the realm of education as well, and the medical schools, and the role of medical schools and the GMC in setting curricula, let alone all the other clinical professionals who need to be involved. No part of the NHS is likely to be exempt from genetics over time. We will need to manage the expectations of the public and healthcare professionals to ensure that this is developed steadily rather than, as the media might wish, “Oh, there’s a cure, I want it tomorrow,” without working through the safety and the cost-effectiveness in taking these things forward. We must make sure that our patients are offered genetics and genetic technologies in a way that they understand and it is helpful to them in managing their diseases. We will need to address ethical and societal concerns about these new technological developments. Mark will be able to talk to you about the Human Genetics Commission. Clearly getting the timing right will be important. I was interested that you, with the expertise of your advisers, suggested that this will come in five years. I think it will be very much two steps forward, three back, five steps forward, two back. It will be some in five years, but it will be a decade before we see a lot of this really paying off for our public and patients in improved care.

Q62 Chairman: Do you feel government policies just now are right for this development, which could occur at a pace? Do you think policies are in place that can deliver advances that come?

Dr Bale: My Lord Chairman, in terms of how the NHS will cater for patients and how it will make use of the knowledge that is coming out from the research, we feel the policies that have been in place since 2001, set out in the then secretary of state’s speech and written in more

detail in the White Paper in 2003, are broadly right. We have recently completed a review of the White Paper.

Q63 Chairman: We thought the review was quite timely in relation to this inquiry.

Dr Bale: Absolutely. We mainly heard – the take-home message – that it is too soon to say this is being done and completed, and, in fact, we need more of the same. We have taken that message on board through the review, in extending some of the key initiatives that we have put in place in the White Paper, particularly the Education and Development Centre in Birmingham, the two reference labs in Salisbury and Manchester, the UK Genetic Testing Network, and other activities in addition to those as a way of helping to co-ordinate, evaluate, and do tests. Policies that we have had in place since 2001 seem to be broadly fit for purpose, and, as Sally said, we look forward to the findings of the review as to what more can be done.

Q64 Lord Warner: Could I pursue that response. One of the issues which came out very clearly for me at our seminar was the extent to which there was a tension between the rapidity of knowledge in this area and the need for some degree of central strategy and direction, and a culture of management and operations in the NHS, to devolve a lot of responsibility down the line in terms of delivery of care. I am just wondering how you are going to reconcile these two. In the seminar there was quite a lot of anxiety that they would not be reconciled very well.

Dr Bale: I think you are absolutely right, there is a tension between the pace at which the science is developing and the initial associations between genes and disease and the time it takes to properly evaluate their use, both in the National Health Service laboratories and also in clinical practice. Those evaluation steps do take time and do take a lot of data to be built up. We have acknowledged in the review recently that there is an issue around how to ensure that commissioners and commissioning can cater for the new tests, which may have different

approaches from the way you have managed certain sub-sets of the population. Your point is well taken and we will be looking at the commissioning angle.

Professor Davies: Perhaps I could add that we had a generous CSR settlement for research. It is now ring-fenced for the NIHR and we have increased the amount of money available for evaluating tests and medical advices, so there should be much more money available for doing those cost-effectiveness studies.

Q65 Lord Winston: How much of that money will be spent on, broadly speaking, genomics and who controls that?

Professor Davies: The budget will rise to £5 million per year. The amount spent on genomics will depend on the applications because we only fund good quality applications.

Q66 Lord Winston: In response mode.

Professor Davies: Initially in response mode. We reserve the right to do some calls if we see something and our policy colleagues want us to on specific areas such as genomics.

Q67 Lord Winston: One of the issues about all these “omics” we are getting into is that there is clearly progress being made, particularly in the field of cancer and so on, but it is absolutely obvious that there is going to be a need for a huge amount of computing hardware, and we are interested to know how that will be set up and how long it will take roughly? How much do you think it is going to cost to do? Who is going to evaluate what you are doing in the genomic area? That is part of my question, but there is another part to this. You have given a very gung-ho view, which clearly my Chairman has led you to give, which is the notion that genomics is the happy future of medicine, but it is quite a determinist view and it is possible that we may find increasingly that epigenetic effects are much more important than we hitherto imagined and, therefore, issues about gene expression and about proteins will

become increasingly important, not the printing of the DNA. Your computing simply looking at DNA sequences is not going to be particularly valuable, I suspect. I wonder if you could lump all that together in some sort of answer. I am sorry it is such a complicated question but they are linked issues.

Professor Davies: Lord Winston, let me start from the back. When I talk about genomics I include the epigenetics and the profiling. I do not think you can separate them. There is ample evidence that you can screen for types of breast cancer by looking at the expression profile. You can look for inherited breast cancer from looking for the genes. Clearly we are talking about it in its broadest sense. Let us address the computing. I think we have to separate two issues. First, what is the computing power needed for the research that is being done that will tell us how we should go forward? That is immense. From the Department we do not have a very big contribution to that. We are very lucky in this country, as you know, with the Wellcome Trust and the Medical Research Councils. At the moment, my understanding is that we have a sufficiency for the grants that the Wellcome Trust and the MRC are putting out. The issue is hardware, but the Wellcome Trust at Sanger have just invested vastly in new hardware, and the issue then, when we know that something needs to be taken through from the research laboratory into the NHS, becomes: Will it need such complex hardware and software? Frankly, if it needs a massive complexity, I think it is going to be very difficult for the everyday commissions to use. We have to find ways to make it simple, where the algorithm is embedded in the software, and that is a research task. We have to look at the task and judge what is needed for that task. Delivering health care will be a different task from researching what should then be transferred across, I would submit.

Q68 Lord Winston: How will you evaluate your progress? Who is going to do the evaluation?

Professor Davies: The review of the genetics White Paper was a good start in evaluating progress. That is not, I should say, because of this review. It was always planned to occur at that time. Do we have any other?

Mr Williams: We have some validation programmes in place for the stem development project that is being evaluated by Nottingham University, but that is a fairly small element of the White Paper spend. We are evaluating the other projects, such as pharmacogenetics.

Q69 Lord Winston: Is it your intention to have a blanket programme, or do you think it might be better to focus in specific areas; for example, diabetes, or heart disease, or whatever it might be that you feel is uppermost in the health care area?

Professor Davies: I have never thought personally that one would go for such a targeted programme, because things come through at different stages. We already know that a number of genetic tests are in use. As a haematologist, I have used genetic tests for many years. As a sickle cell doctor, I have used genetics in my everyday work. If there were a cost-effective genetic approach, to deny it to the public would go against the philosophy of the NHS.

Q70 Baroness O'Neill of Bengarve: Could I push you a little bit on cost-effective. When we come to single-gene disorders and relatively rare, is it reasonable to concentrate on those that will benefit rather few people as against those that might benefit a very large population; for example, something relevant to diabetes? Is there not a choice here?

Ms Payne: The developments around single-gene disorders have been significant and we already have a large number of genetic tests available. I know that if you spoke to, for example, Alastair Kent of the Communities Interest Group, he would make the point that, while these conditions are individually rare, the proportion of the population that is affected by rare disorders as a whole is quite substantial. If we are concerned about anything in the NHS, we should be concerned about making sure that people receive care on the basis of their

need. It is not an either/or situation. We do have special genetic services which can meet the needs of those people and we have a system in place to make sure that we use genetic tests and services in a cost-effective way. We only use a genetic test for an individual with a rare genetic disorder if it will inform their clinical management and, therefore, we expect that we are using it in a cost-effective way. That does not mean that we cannot, as developments pull through, extend the use of genetic information to broader areas, be it sub-sets of common disorders, as we currently see, for example, in breast cancer, but also as we start to get more information about the genetics of complex common diseases.

Baroness O'Neill of Bengarve: If I may, my Lord Chairman, I will park this question, because it is one about cost-effectiveness and, in a sense, can probably be answered more readily when we are further into this session.

Chairman: Thank you.

Q71 Baroness Perry of Southwark: Your two departments have jointly set up the Office for Strategic Co-ordination of Health Research. In relation to genetics, what do you expect to come out of their work? How do you see it translating the basic research into something which will help in the clinical field?

Professor Davies: The OSCHR is a small joint office of the two departments, hosted by the Department of Health, that has an independent chair and three non-executive board members, plus the Chief Executive Officer of the MRC, the Director-General of Research through DIUS, myself, and my deputy in lieu of Chief Executive Officer of the NIHR. The objective of OSCHR is to make sure that the two delivery agents of research, Medical Research Council and NIHR, co-ordinate, so that we can get pull-through and push-through, so that both delivery agents are focusing on strengths. We set up a translation on the Medicine Board that looked at how it is looking, and how one moves from fundamental research through to targeted development, through to the early studies in man. All of those and experimental

medicine, the MRC leads on, and then, as it gets to phase 2B studies onwards, including, finally, the health technology assessments, we, through the NIHR, lead on that. The role of OSCHR is to ensure that we are interlinking to deliver pull-through and push-through. Does it have a specific role in genomics? That board suggests to us areas that we do need to be cognisant of, and so we are doing work in public health. We both do quite a lot of work in genomics, as you are hearing and will hear this morning. I would be surprised, because of its pervasiveness, using the broad description I have already accepted, if we did anything more targeted, because it is so pervasive in everything that people find.

Q72 Baroness Perry of Southwark: You mentioned the membership of OSCHR. What relationship does it have to BBSRC and to BSRC? How do you communicate with them?

Professor Davies: There are two relationships. One is the Director-General of Research from DIUS. The science vote comes under him, and the officials work closely with Paul and he can talk about that. Of course, Sir Leszek Borysiewicz as Chief Executive of the MRC is part of RCUK and therefore links into them like that.

Mr Williams: Perhaps I can expand a little bit. One of the issues that came up when the Cooksey Report was received was how far the research activities of the councils other than the MRC would be fed into the OSCHR process. Sir David Cooksey said that he did not favour taking away responsibilities from those councils. We already have quite good co-ordination of members between the seven Research Councils under the umbrella of RCUK, so this we regard as the main mechanism for ensuring that there is co-ordination at the highest level between the councils. In reality, those councils have co-operated in areas of mutual interest with both the relevant managers of the MRC and also with colleagues within the Department of Health and NIHR. That might seem a slightly untidy relationship, but the difficulty is that we wanted to ensure that the OSCHR arrangements did not pull the MRC away from the Research Councils, precisely for that co-ordination. It is something we are

aware of. It was a risk that that could happen if we were not careful. So far, however, particularly if you take the ESRC, which has really become very active in the OSCHR's responsive field of activities, I think that mechanism seems to be delivering.

Q73 Lord Warner: Could I pursue how this relates to the big bad world of operational delivery. There is another universe outside research: it is the Department of Health's planning and budgeting system/priority settings/operational framework which the NHS uses to decide what it is going to do, how it is going to spend its money, and what its priorities will be. I am not clear how all this research panoply of co-ordination, which is set out in your paper and is mentioned there, fits with that other universe. Or it is a parallel universe which does not meet up with the research universe?

Professor Davies: I sincerely hope it is not a parallel universe. Clearly there is an issue of prioritisation. Is research going to be useful and matter? Most of these programmes that I have been talking about for NIHR are being run out of the NIHR Evaluation Trials and Studies Coordinating Centre in Southampton – which, I believe, Lord Warner, you visited in a former incarnation. We have down there a very big prioritisation process. It involves web prioritisation from clinicians and patients but, also, it involves managers from committees and patients. So there is a prioritisation process, a priori; meanwhile, I have committed £3 million a year to the top priorities of NICE and we have the opportunity for other key stakeholders in the policy arena to put in priorities. We try to make sure that important priorities are addressed. Before anything is commissioned, it goes in front of a panel of lay people, service managers from the NHS, as well as academics. It must be methodologically good but the issue as to relevance is also part of our expert review.

Q74 Lord Taverne: The White Paper has been successful in establishing procedures and facilities for single-gene disorders, but what about the more common human diseases? How

successful has the White Paper been in widening genetics to mainstream specialties? What progress is being made in establishing procedures, facilities, and expertise in that case?

Ms Payne: I think it is fair to say that the mainstreaming agenda has inevitably been rather more challenging. Under the genetics White Paper we have funded a number of different kinds of pilot projects which have looked at taking genetics into mainstream clinical specialties – in the current state of knowledge, generally still dealing with single-gene disorders but in things that particularly are related to renal disease, for example, or to certain cardiac conditions – and tried to develop a more patient-centred service which integrates genetics knowledge, genetic counselling, and elements of a specialist service, with the relevant clinical specialty knowledge about how the condition will then be managed in all patients once it has been clinically diagnosed. We have done a number of different kinds of pilot, some in a joint programme with Macmillan around cancer services, and a number of different ones around different clinical specialities, such as renal and ophthalmology. We have also had some pilots looking at putting GPs with a special interest in genetics into primary care across ten PCTs. That is not about doing a clinical service but about providing a resource to primary care about understanding genetics and the implications for the care of patients in primary care. Another pilot looked at familial hypercholesterolaemia, to identify people at risk of that condition through cascade testing from cases already known. All of those projects either have completed or are in the process of completing their own evaluation reports, highlighting the challenges they faced and the barriers, the lessons learned, the successes, and some of the things we will need to look at in the future if we want to develop these kinds of services further. To complement that, as my colleague Mark has mentioned, Nottingham University is doing an external evaluation of all service development projects. That is looking much more at the kinds of issues that perhaps Lord Warner touched on, the organisational issues of how to get these things embedded in the NHS and some of the

challenges about getting NHS organisations to understand how to implement and commission slightly different kinds of service. They will be reporting in the autumn of this year. Once we have all of those lessons, we will be looking at how we can share some of those lessons with the NHS and at areas which may have been highlighted where further action may need to be taken. Although that is not quite the same thing as genetics in complex disorders, where we do not have very much that is relevant to clinical implementation yet, it will throw up a lot of lessons that will be common to that, which we will be able to use to develop services in the future and to look at what kind of infrastructure we need in place.

Q75 Lord Taverne: How much funding is going to go into this development?

Ms Payne: Into the pilot projects?

Q76 Lord Taverne: Into the more common diseases as opposed to the single-gene disorders.

Ms Payne: I do not have the figures on me at the moment about the money going into the pilot projects. They were not complex diseases, in the sense of the genetics of complex disease; they were single-gene disorders relevant to these particular clinical specialties. They are not quite the thing that you are talking about. In fact we do not have areas of clinical implementation where we are dealing with the genetics of complex disease, because we do not yet have the evidence base for how we are going to use that in clinical practice. I am not an expert, but I suspect that that is one of the things that is probably more than five years hence, possibly more like ten, although there may be individual examples before that.

Dr Bale: It is important we understand what we are considering here. My simplistic approach to this is that we have common diseases where there is a rare genetic sub-set which is caused by single-gene disorders. We have seen numbers of those. The BRCA1 and 2 genes in breast cancer is a good example. Those are the ones that we have tackled in some of

the pilot projects we have done to date. We also have some projects targeted on the very rare single-gene disorders that Baroness O'Neill mentioned. We are not yet tackling ourselves – we are certainly looking for a way forward on this – where there are multi-factorial genetic mutations or polymorphisms, which are implicit in development-related disease. That is a much more complex area. That is where the scientists are still developing the answers and we do have to look very carefully at how that knowledge will be transferred into clinical practice. That is a very big challenge for us. We acknowledge that.

Professor Davies: Under NIHR we have funded a series of Biomedical Research Centres and Biomedical Research Units. We are spending £100 million a year on the centres and £12 million a year on the units. Most of them have genetics themes within them specifically, as genetics themes, but, also, genetics in all the other bits. Cardiovascular in Oxford has a big genetics component; diabetes and cardiovascular in Cambridge have a big genetics component. Those centres are specifically to bring the results of research, pulling them through so they make a difference to patients. That funding will help make some of the more complex multi-gene disorders real for patients and what we need to do. But surely a lot of this is about education as well. Education starts in the schools and there is no doubt that children are being educated in school in a way that they were not before. I only have to look at my 13 year-old and 16 year-old. My 13-year old told me this weekend that there was clearly a gene for collecting polythene bags and storing excessive numbers that went down the maternal line in our household. They are learning in a way that we did not – the knowledge was not there. Medical schools are teaching them. Now I find that the young doctors coming through understand much more than they ever did previously, and so I have a hope that that will take us forwards.

Q77 Lord Colwyn: I must apologise for my late arrival: I had to see some patients this morning and I was over-running, hence my delay. The genetics White Paper envisaged the

NHS leading the world and taking maximum advantage of the application of the new genetic knowledge for the benefit of all patients. Given the possible beneficial and detrimental impacts of genetic tests, how are the Government going to evaluate the effects of genomic tests upon diseases?

Professor Davies: We would like Britain to be at the forefront of doing things well and right. As I explained a little earlier, as these tests become available we do have the capability for commissioning evaluations either responsively or as a commissioned project through from our centre of the NIHR in Southampton. I envisage we will be spending a budget of up to £5 million a year evaluating tests and devices. We have set up a specific programme with funding.

Dr Bale: In the UK we have the UK Genetic Testing Network which was set up to provide a consensus-building, co-ordinated arrangement for the NHS genetics laboratories. It has pioneered the gene dossier approach to looking at the evaluation of genetic tests, both clinical and end-of-evaluation. It requests that laboratories compile a dossier for a test before they are provided to the network, so they are evaluated. Although it is a very simple model, it is the envy of a number of countries in the world who look to the UK for leadership. We are quite proud of that. We are also very pleased that they are beginning to look to the future. They have recently discussed a document prepared by academics in Cambridge around the evaluation of more complex diseases and biomarkers. This is now being sent more widely to the professionals for their views on how the UKGTN model might be adapted for these more complex diseases. When they have the results back from the informal consultation, we will look forward to hearing from them as to what they think the UKGTN might do. I believe they will then be looking to apply this new approach to one or two modern diseases that might be provided by laboratories. I think the approach taken by the UKGTN is applicable more

broadly. I certainly know that it has been looked at with interest by a number of international bodies like the OECD and workshops in the UK about evaluation of genetic testing.

Professor Davies: Speaking as a haematologist, we should not forget that our pathology laboratories have undergone many changes in the way they do things over time. They are beginning to use genetic tests in routine pathology laboratories and have established mechanisms for evaluating tests for use in the NHS as well.

Q78 Lord Colwyn: How are the Government going to ensure that healthcare professionals have training to deliver this advice? Also, how are the needs of professionals who are not genetic specialists going to be met? How is this being achieved in practice? Where is the funding going to come from?

Ms Payne: We have already referred to the work of the National Genetics Education Development Centre. We see this as having an absolutely key role here. It was set up under the genetics White Paper, not to deliver the training Directive itself but to work with the established regulatory professional and academic bodies that are responsible for delivering pre-registration/post-registration training and continued professional development to make sure that appropriate levels of genetic knowledge are incorporated into the relevant curricula and training and, also, to help them identify the needs of the various professional groups, which obviously may differ from group to group, to help source resources to support that and to develop some resources themselves. The initial phase of establishing the way they were going to work took a little bit of time, in developing those networks and links to the relevant bodies, but they have now started to do some really valuable work in taking forward this specific agenda. For example, they would work with the Royal College of GPs to make sure that there was now a statement about genetic competence within the curricula for newly qualified GPs. They have done some work with dieticians to identify what their needs might be and what kind of genetic knowledge they might need to deliver their services. They have

recently – I think only in the last few weeks – published a report about pharmacy and the kind of knowledge that pharmacists would need in relation to the developing world of pharmacogenetics and the different kinds of knowledge they might need to implement those things. They have done some generic developments around core competencies which will help support new kinds of roles in the NHS, where they will pick out relevant competencies that are needed for particular jobs in order to identify the skills individuals will need. They have also developed a really powerful learning tool, a resource called Telling Stories, which is a database of patients explaining, sometimes with video clips and sometimes with sound clips, their experiences of using the NHS, and then using that to highlight some of the issues that person needed to address in relation to the genetics of their disorder. Because of the current state of knowledge, that tends to focus on some of the single-gene disorders, but they are already looking at how they can expand that to cover a wider range of diseases, so that it is relevant to more and more clinical specialties. We feel that in the education centre we have established a really valuable resource for supporting this. Professor Farndon, who heads the centre, has already submitted evidence to the Committee. One of the key things he would highlight is getting the right moment when you approach particular clinical specialties. If you go when there is not yet anything of particular relevance to them, you will easily get disengagement and they will say, “This is not relevant to us.” Timing is everything. It is getting the right moment to go into a specialty. When they can see there is a relevance, the experience of his team is that clinical specialties are very keen to engage and look at how they need to incorporate genetics into future developments.

Q79 Lord Colwyn: Tomorrow we are debating allergy in this House, and there is no money to pay for centres for allergy treatment. Are there going to be problems with funding in this new work?

Ms Payne: This is about incorporating genetics into the knowledge of the workforce. The workforce will be being trained or will be having continued professional development and this will be an element of it. I do not think there is necessarily going to be huge additional expense involved in making sure this is appropriately incorporated into the curricula. This is part of the everyday job of these professional regulatory bodies. It is just making sure that we support them in knowing what they need to address and include and how they can tailor that to the needs of their particular professional group.

Q80 Chairman: Apart from the areas that you mention from the centre about education, we have not had any evidence from any of the professional organisations which said they are involved in education and genetics. Do you not think that is a major gap?

Professor Davies: Then their education is probably failing, because most of them ought to be aware of what is coming. I have not looked at who you have had evidence from.

Q81 Chairman: Maybe it will come. I do not want to exaggerate.

Professor Davies: We are trying to mainstream genetics into education because it will be fundamental, in its broadest definition, to a vast amount of the work of the NHS in the future, particularly as we move towards more preventive issues.

Q82 Chairman: You mentioned pathology labs. If some of the evidence presented to us suggested that the current model of laboratories (pathology; what I used to call bacteriology, or microbiology; haematology; et cetera) and every Trust in the country having the same model replicated is probably out of date when it comes to dealing with genetics and genomics, would you have a comment on that?

Professor Davies: I think that is likely to be true. If you look at how we have developed regional genetics services, it is because we need to aggregate them. However, if you look

back at how pathology laboratories have developed, they were all very separate, and then more and more they have come together as departments of laboratory medicine because the technologies were common, so you could afford the technologies and the robotics once, in the hospital, and different specialties used it for different tests. That may be one of the futures for genomics, in part – at least in big conglomerations and big hospitals – but we do have, as you know, a second phase of the pathology review going on by a noble member of your House, so he may well have views about this.

Q83 Lord Warner: I think this is an area which we will need to pursue as a committee. We will need to have our own contacts with the people responsible in this area, but I would just like to be clear from the research perspective. With the advancing knowledge in the range of tests that will have to be performed to get the greatest benefits out of discoveries in genomic medicine, is it the view that there will have to be a considerable consolidation of pathology department expertise if we are to get the maximum benefit?

Professor Davies: I think that is likely to follow if you look at how research genomics has gone. More and more, rather than set it up in your university's back yard, people are getting grants and saying, "So who can supply this many snips on that chip at the cheapest price?" and, indeed, with the dollar going down, buying in from the States. The evidence from the research is that more and more people are doing factory type work. If that holds true, it may well be that we have to look at that for the NHS.

Q84 Lord Warner: Perhaps I could move us on to the issue of regulation. The UK Genetic Testing Network regulates the provision of genetic tests for single-gene disorders. Given the increase in the scope of genetic testing, both within the NHS and, indeed, in direct-to-public tests, do you think the current regulatory framework is sufficient? If not, what are the kinds of changes you would expect to see?

Dr Bale: We do not consider the UK Genetic Testing Network being able to regulate genetic testing in the NHS. It provides a forum for consensus building and evaluation for testing and then makes a recommendation to the NHS commissioners as to which tests to commission in their areas. It also provides a network to allow particularly rare diseases to be concentrated in one laboratory rather than being done by a number of laboratories at a very slow pace. The UKGTN is a model for evaluating, primarily. When they have evaluated a test – they base their evaluation on evidence from the Health Technology Assessment Programme and so on – those tests are then recommended to the NHS. They do not have a role at all in evaluating tests provided directly to the public by commercial providers. There are some regulations, which stem from the EURO Directives on Medical Devices, that regulate the sale of genetic test kits which are sold to the laboratories and sold to the public, if there are such kits. They are overseen by the Medicines and Healthcare products Regulatory Agency and they have very close liaison with us about these aspects. The Human Genetics Commission has established that there is a gulf between the regulation of tests and services provided directly to the consumer and the situation we have with the MHRA. They have looked at this again recently and recommended to ministers that there should be some thought given to a voluntary code of practice to try to regulate the commercial providers of these tests. It is an option that the ministers have asked the HGC to go back and support further with the industry, because we find that the industry themselves are concerned about the variation in standards. This largely comes down to issues around claims made for the tests: the advertising, the marketing, the public expectation which can be built up for some of the tests. There are also issues around privacy, confidentiality, and consent. There is an offence under the Human Tissue Act of testing someone's DNA without their consent, which is an important safeguard. The HGC will be talking to the industry. They are not meeting with them until June and then we will be seeing if there is appetite for a code of practice and what

the scope of that code of practice might look like. We certainly have models from other areas, such as paternity testing, which seem to be successful, but putting this into an authoritative document does raise standards.

Q85 Lord Warner: I agree I was slightly misleading in what I was saying about the work of the network, but the reality is that there is often going to be a blurring in the area between evaluation and regulation. At the moment there have been a relatively containable number of tests which you are dealing with in relation to the NHS, but, from the evidence we were given in our seminar, with the explosion in knowledge in this area it would be astonishing if you did not see a huge step-like change in the number of tests that are available in one way or another, good, bad, or indifferent. We are trying to probe a bit more in relation to how, whether you call it evaluation or whether you call it regulation, you cope with that volume. The line between direct-to-the-public and NHS use will get blurred in the public's mind. Because it is something that is available, many of them will assume it is something which is available in the NHS: "If it's available in Boots, why isn't it available in the NHS?" I am trying to understand what preparation the Department may be doing or the Government may be doing to cope with the volume increase of a probable scale.

Dr Bale: We would accept the role of the UKGTN. We have certainly made investments since the White Paper to increase the capacity of laboratories that are in the genetics network. They are now able to cater for much larger numbers of tests and turn them around towards the times that were set out in the White Paper. When it comes to the broader role of genomics in pathology services, that is another conversation that is being had within the Carter Review. The important aspects that we have to recognise for some of the tests – and it is not just evaluation, you are absolutely right – are around quality assurance and the ability to have faith in the results that are coming out of the tests, particularly in the very important ones. Certainly the UK is amongst the leaders in the science and quality assurance and external

quality controls and so on. One of the UK scientists, who is a former Head of the Reference Laboratory Manchester, led some work by the Organisation for Economic Co-operation and Development to produce international guidelines on this, so we are aware of those aspects, as well, about the need to have faith in laboratory tests. The funding for the laboratories and the funding for the tests coming through will have to be catered for in the normal commissioning arrangements. Those are undergoing a considerable evolution at the moment, but, as the tests come on stream, as the evidence base is analysed as robust, and as the profession is saying, “This is the test that we need to be able to supply,” the commissioning process will have to be able to address that.

Q86 Baroness O’Neill of Bengarve: Perhaps I could take you back to Genetics Knowledge Parks, if I may. We have one in Cambridge which has produced a very interesting mix of work. I have kept in quite close touch with them. You have submitted that they did make a significant impact in some ways. Then, of course, the funding was discontinued. That may be exactly the right decision, but I would be very interested to understand more about why the decision was made at that stage to close down one route to embedding genetics in mainstream medicine. Was it that it had achieved all it could achieve? Was it that some other route to the same end seemed better? Or was it simply a funding decision, where some things had to go?

Professor Davies: Probably – as all these things are – a mix of many of those bits. We too would say that the Genetics Knowledge Parks were a success. Having been with them – as was Mark, who will add to this – from their inception, they were intended as five years and then to be self-funding, and the DTI was a co-funder at that time. We found, as they developed, that the whole field began to take off and a number of other people were working in those areas. The ESRC is now funding increasing amounts of the ethics and social areas. The science was interesting but, under Best Research for Best Health, the Government strategy for the NHS’s science, we are putting in rather more money available to everyone

rather than just the five Genetics Knowledge Parks, so it was an opportunity then on merit for everyone to be able to work on genetics.

Dr Bale: That is broadly an outline of where we stand. We have already mentioned the education aspect. They did have a role in both educating and helping to educate clinicians, which we now see as being primarily a role for the National Educational Centre to lead on. The public engagement angle that they tagged is now largely funded by Sciencewise, which is running within DIUS. It is really a question of recognising the pioneering role they had, and they are now seeing that the funding is being picked up. Many of the same individuals are operating through the ESRC centres, the Biomedical Research Centres, and so on. It is an idea that was mainstreamed itself into the wider research community.

Professor Davies: I should say that we are about to set up under the NIHR Networks for England, research networks, a study group on genetics. They will be able to help ensure that the genetics studies that are funded by ourselves and our partners are effectively undertaken through the research networks in the NHS. They will be able to help each other to develop a portfolio of genetics applications to the funders, including ourselves, and we will be able to feed in some comments about the research arena and NIHR.

Q87 Lord Winston: You mentioned Best Research for Best Health. One of the things that was pointed out is that you are intending to create a health system in which the NHS supports outstanding individuals in world-class accommodation. How are you going to implement that?

Professor Davies: I think we have made an awfully good start. We not only have an increased budget through the Comprehensive Spending Review but for the first time ever we have a capital budget, £50 million a year. That, for last year and a significant amount of this year, is being distributed to those who have, through international peer review, won our centres and units grants. Some of that, I am sure, is funding genetics equipment for research

near to patient and with patients in our major centres. You will know about the Biomedical Research Centres. The Biomedical Research Units we announced about a month ago, and they are in areas of major health need. They are dotted around the country, building on small teams that are very good, many of them doing genetic work. We are giving them capital and helping them. We are making a good start, but one has to remember that this is a three-legged stool: the NHS is making its contribution; academia, from HEFCE-funding through the universities, makes its contribution; and the Research Councils make their contribution. They all come together to provide the greater part, as it were. That then takes you into: Do we have the right model? You will know that Lord Darzi in his London Review highlighted the role of Academic Health Sciences Centres as a way of driving innovation. I can only think that genomics, as well as robotic surgery and other things, must have been behind some of that thinking. I do not seem to be able to go anywhere these days without being asked: “What’s the Government’s view about Academic Health Sciences Centres?” – for which one has to wait until his next review is out in July – and everyone wanting to do it, because they seem to see this agglomeration and coming together of academia and the Health Service as a way to really make it work better by the Better Results structure.

Lord Winston: So you are going to continue to support Imperial College!

Q88 Chairman: You do not have to answer that.

Professor Davies: I support Imperial when they win in competition.

Q89 Lord Winston: You do not need to answer that – and I would declare an interest if you did. I am interested in an answer that Mark Bale gave about Sciencewise. One of the issues with regard to world-class research by world-class individuals in this area is the need, particularly in the field of genetics, to support good animal research. That is becoming increasingly difficult, particularly in the genetic arena at the moment, because of increasing

regulation and demands that are made which tend to turn a lot of young people who might do PhD projects away from that kind of work to something which is a bit easier and will not require such a lead time to get research approval and a Home Office licence. I am interested to know in which dialogue processes with regard to genetics and the use of animals you are involving the public. This seems to me to be an increasingly important area.

Dr Bale: I am afraid I am not aware of any specific public dialogue involving genetics and animals. I do not know if colleagues from DIUS have any knowledge of that. Sciencewise I think tend to operate in a response mode, so when topics like that are suggested they can fund them. I would have to check on the specifics of that. I certainly know the Academy of Medical Sciences has expressed an interest in doing work, in parallel to the work they did on human and hybrid embryos, to look at what might be done in animal hybrid embryos, which would include potentially genetically modified animals, animal models, and so on, but as far as I am aware that has not been taken forward yet.

Q90 Lord Winston: I do know that on the Sciencewise agenda animals was one of the possible areas they might look at. I think it would be extremely helpful if the Department of Health took an active interest in that.

Professor Davies: We clearly do. There is a ministerial group that meets about animal research and it is presumably led by DIUS. I do not know.

Dr Bale: I think it may be Home Office.

Professor Davies: Our policy is that we support animal research and its essentialness. We do not fund it, because we fund around patients, but we do support it.

Lord Winston: It is not the funding that is the issue, it is the information and the response to the information, and the need, which is becoming increasingly clear cross-government, for dialogue in all sorts of areas. This seems to be a very important area.

Lord Taverne: Perhaps I could follow this up. Could you take due note of Lord Winston's remarks and make representation to the Home Office because the House of Lords Committee on Animal Procedures and Scientific Research made several recommendations, most of which have been followed up very well, except for protests about the bureaucracy involved in the rules that the Home Office still insists on in animal research. It is oppressively bureaucratic and very little progress has been made in that field.

Lord Winston: Particularly in the genetic area, which is what I am focusing on, of course.

Q91 Lord Taverne: Would you please make representation to DIUS?

Professor Davies: I will happily pass this on to them, yes. The other bit which I did not pick up on earlier, Lord Winston, is about how we make it first class. That is of course our new training schemes to develop clinical academics. As you know, we will be rising to 750 clinical academic fellows and 400 clinical lecturers, many of whom will be working in the genomic field. We also have money for an extra 25 medical PhDs – we can claim eight a year, so that we have a rolling programme. At the moment, nine of the academic clinical fellows supporting 19 posts and seven of the clinical lecturer programmes supporting ten posts are in genetics, so there is a significant academic cohort coming through.

Q92 Lord Winston: Are these research studentships funded through the R&D budget or through the Research Councils?

Professor Davies: I am talking about the NIHR budget. The MRC funds a large number of people in genomics and some bioinformatics. It always has done.

Q93 Lord Winston: We have had nothing at all so far from the TSB about some of these issues. It would be extraordinarily interesting and useful to know where you interface with all these strategic issues.

Dr Manimaaran: The TSB works very closely with the Department of Health and the Research Councils in trying to define an agenda where it intervenes. It works very much closer to market in terms of the near market technologies. In an area like this, it would look to work with industry to see what kind of market opportunity there is for UK capability, particularly industrial capability, and then particularly with the Department of Health, which own the policy agenda, and the Research Councils in terms of translating that kind of basic science capability to technologies in the market. They are developing their strategy in health care and bioscience at the moment.

Q94 Lord Taverne: Can I follow that up and ask about the interface with this and the fact that the DIUS and the National Innovation Centre have a cross-government role in innovation. Do they have a specific strategy for genomic medicine?

Professor Davies: The first is working with the Technology Strategy Board. Each research funder works with the Technology Strategy Board. The Department of Health is one of two departments piloting their new ways forward. We have not yet, I think, decided which areas, although the first platform we launched with them was on assisted living, and we are looking at what the areas will do. The MRC is also working with them and we co-ordinate with the MRC. On genomics, there may be things that come up, and the Technology Strategy Board is clearly building on the link work – a lot of which was with SMEs and in the genomics field, so I would think they will. The National Innovation Centre is part of the National Institute for Innovation and Improvement and it was set up as a result of the Healthcare Industries Taskforce – the hated taskforce – to help people navigate the innovation landscape – which is extraordinarily complex, particularly SMEs and the Medical Devices people. The National Innovation Centre has no formal role relating to genomics; it is about devices, diagnostics. They are helping smooth the path and they co-ordinate the NHS innovation hubs as well.

Dr Manimaaran: In terms of DIUS, we are the policy across government in terms of fostering innovation, but there is not a specific role on genomic medicine. That is clearly a Department of Health area. We do fund the Technology Strategy Board, and we provide the framework within which the TSB undertakes its activities along the same lines as the Research Councils. We provide them with strategic thrusts and encourage them to work with the Department of Health and work with the Research Councils in co-ordinating healthcare research and providing funding where they see a specific opportunity. But DIUS itself does not have an innovation policy relevant to genomic medicine.

Q95 Lord Taverne: Has the TSB supported particular projects in genetic projects?

Dr Manimaaran: I believe it has funded a handful of projects to date through what have been specific calls for healthcare research. They have had some R&D projects focused on biomarker discovery and translation. It also has a couple of initiatives that are relevant but not completely related to genomic medicine, such as Stem Cells for Safer Medicine, of which it is a partner, and a pilot Healthcare Technology Co-operative, but it has not done a great deal of work on genomic medicine to date.

Q96 Lord Taverne: Are there any special plans for the long-term future, beyond the next three years?

Dr Manimaaran: Early this year it got its comprehensive review settlement and it is developing a strategy. It has developed its overarching strategy, which is being published on 8 May, tomorrow, and in the case of specific technology areas it will be publishing strategies as well. But this is clearly an area where it will work very closely with the Department of Health and the Research Councils.

Q97 Baroness Perry of Southwark: We are still trying to wrestle with these various bodies and how they relate to each other and exactly what they do. As I understand it, TSB is very much emphasising the link between research and implementation. Does it talk to OSCHR? Given that OSCHR has distanced itself from the application into clinical practice, is there an area in genomics where these two are talking to each other? Should they be talking to each other?

Professor Davies: I can tell you that they have met, although I do not know what they have discussed, but, as OSCHR does not have a budget (the budget belongs to the MRC having come through DIUS or to NIHR having come through the Department of Health), their role is not to set the strategy. There are governance mechanisms for the MRC and NIHR to ensure we co-ordinate and push through and pull through the full spectrum of work, and perhaps to highlight an area like public health if it is considered the nation has not focused enough on it.

Q98 Baroness Perry of Southwark: Surely, if OSCHR is there to do a strategic co-ordination, you have said it has met once, should it not also be interested strategically in what the TSB is doing?

Professor Davies: I am sure it is, but the Department and NIHR are interacting with TSB – the interaction needs money and we have the budget – and the MRC is working with TSB, and we work together and we tell OSCHR what we are doing.

Q99 Lord Warner: Talking of budgets, in paragraph 23 there is an interesting paragraph about the Innovative Medicines Initiative, which is, as I understand it, a private/public partnership with quite juicy sums of money available over the next ten years: €1 billion from the European fund and a matching €1 billion from the private sector and industry. Could you tell us a little bit about how genomics and pharmacogenomics will be included in this

initiative? To what extent are the Government involved in the decision-making process and how will they be extracting benefit for UK plc from this initiative?

Mr Williams: IMI is a EU Commission initiative, so the UK Government is not formally involved in the decision-making process within the IMI framework, although we have general oversight in relation to the framework of seven funds. The IMI has just issued its first call, at the last end of last month, which covers about 18 themes. A number of those give very clear opportunities for those interested in genomics to come forward with proposals for consideration by IMI and the Committee. There are big opportunities there for people in the UK to seek that funding and to deploy it.

Professor Davies: The Medical Research Council did help groups, before the call came out, to get together to think about what sort of thing they could put in and everything, so I can tell you from my academic links that there is quite a buzz out there of people starting to network.

Q100 Lord Warner: Is there a Government enthusiasm for targeting a big UK benefit from this? Could you tell us a bit more, not so much from Sally's academic position, but about what the Government are doing to drive a little bit the UK take up in this call for initiatives.

Mr Williams: We have a number of mechanisms. As Sally said, the MRC has taken the lead in trying to bring together consortia which may be able to benefit, because the consortia that it would deal with if it was on a national basis are precisely the type of organisations that would potentially benefit from IMI. They have led that key role. There is also the UK Research Office in Brussels that provides an information source for people seeking funds from Brussels. This is a joint Research Council initiative, managed for them by the ESRC, which provides access directly to the Commission and subsidiary bodies who advise on how to do that. The way the UK system works in terms of basic research gives this role much more to the Research Councils than DIUS centrally. The money is devolved out to Research Councils

every two or three years. We asked the Research Councils to operate in parallel in relation to the European fund and opportunities of their own.

Professor Davies: Perhaps, Lord Warner, it would be helpful for you to know our past history as a nation: we get out vastly more than we put in from the framework.

Q101 Lord Taverne: I am not pursuing a Euro-sceptic agenda here. I understand what the Department of Health is doing but I am trying to understand, when there is this initiative, given that it is a private/public partnership with industry involved, what the non Department of Health parts of government may be doing to generate a bit of umph in this area for industry; for example, SMEs and so forth.

Professor Davies: BERR have done quite a bit of work with Pharma – who are very keen on this area, as are we – in order to make sure that everyone was aware.

Q102 Chairman: You might also answer how the IMI will publicise the call to industry and academia.

Professor Davies: They do publish through CORDIS and open calls in the *European Journal*. Of course the minute something like this comes out – and everyone was waiting for it: the MRC and ABPI did a joint conference about the fact that it was coming – the emails go spinning around everyone. We were all waiting for it. We all knew it had come. It had all gone around the academic network. If it has not reached someone, then they are not well networked, so they are unlikely to get the funds. Speaking from experience of watching people, you need to write ----

Q103 Chairman: They only get to know through the network?

Professor Davies: No, if they consult the EU website or CORDIS or the *European Journal* it is all laid out, but, in addition, the MRC has been doing work and the academic network is passing it around.

Dr Manimaaran: To expand on Sally's point, the Medical Research Council provides a national contact point service for this particular initiative. You have particularly bodies who are tasked with raising awareness and getting groups together to respond to particular initiatives that come through the Framework Programme, and in this case the Medical Research Council are leading on the provision of the knowledge and awareness.

Q104 Lord Warner: I am still a bit puzzled by this. I do not want to pursue this now but I think it would be helpful to have a fuller response in writing for what the Government has done in promoting this initiative and to have a little bit more about the results of the British applications are in this area.

Professor Davies: We will not know the applications. First of all, the closing date has not happened, and, second, we do not know as a government – even the Medical Research Council will not know – but we will send you a note from DIUS and BERR.

Q105 Chairman: Thank you. In due course, if you get more information too.

Professor Davies: Yes. We might hear some success.

Q106 Baroness O'Neill of Bengarve: The HGC produced a report, *More Genes Direct*, which I am sure you are familiar with, on direct-to-public genetic tests, and there has been a lot of other discussion and concern by other bodies about direct-to-public tests. How are recommendations from the HGC report being taken forward? Do you think this is an area where we are going to need regulation or self-regulation work? Are you concerned about members of the public who buy these tests and find the results incomprehensible to them, or

about the implications of the results then making demands on the NHS for repeat tests with interpretation?

Dr Bale: The HGC has recommended that there should be some work done on a voluntary code of practice. They have also recommended that the issue should be looked at again when the In Vitro Diagnostic Medical Devices Directive is reviewed, and that is something the Government have agreed they will take into the discussions with the European Commission. In the meantime, and given the pace of change in this area and the fact that you may be required to have a flexible response to any services that are provided, the self-regulatory pathway is one that the Commission has been asked to explore further. As I mentioned, they have started to discuss a number of the issues with the various regulators, such as the Advertising Standards Authority, Ofcom, the Human Tissue Authority and the Data Protection Commissioner, and they will be trying to see if there is an appetite there for the setting of standards for the industry themselves. One of the key questions, as ever, with a code of practice, is compliance, NHS compliance. Our experiences with paternity testing have been that once the industry sign up to this they are very good at self-policing. We find that if a company starts to advertise a service in a particularly aggressive way or in a rather snooty way, they are pretty soon shot by their competitors, who have a very close interest in the market. Whether that will happen with genetic testing services is unknown as yet because, so far, there have been so few of them. We have only had one or two fleeting services provided in the UK, although of course it is quite widespread in our Internet and particularly coming from the US. This is what we need to explore with the industry, and the Commission will report back to Government later in the year. Our expectation is that a voluntary code of practice is the right approach at this time.

Q107 Baroness O'Neill of Bengarve: There is an asymmetry with paternity testing, if I may say so, in that paternity testing essentially is going to give you the information that the

source of one sample is or is not very likely to be the father of the source of the other sample, but this is not quite like a test that is taken for health purposes.

Dr Bale: I agree. That issue around the claims made and the information given is at the heart of the genetic testing debate. Many of the companies that provide over-the-counter services or direct-to-the-public services steer very well clear of the single-gene, highly penetrative disorders, those that may have a dramatic impact on a person's health. They look to provide a service which focuses on the weaker associations that might help people to adopt a better diet or maybe to consider the most effective way of stopping smoking or losing weight. Those are the sorts of lifestyle tests that we have seen provided. There have been services provided in the past for carrier testing, if you were considering carrier testing before pregnancy. Again, whether these things are provided privately or publicly is an issue that has not been linked to genetics. Many people go for private testing through the medical profession and other health professions in a way that does not really concern the NHS too much. We need to explore – the Commission has certainly begun a very active discussion on this – what is acceptable and unacceptable to market direct to the public without proper health care support and without proper support.

Q108 Lord Winston: Certainly these are becoming quite big business in places like California. Is there any evidence at all that whilst they might make a difference to health – to “lifestyle” as it is called – they are really effective in what they predict and recommend?

Dr Bale: The jury is still out on that one. This is one of the criticisms of these services: they do not have the sort of in-depth evidence that you require to put this into an NHS service, which is where you will find a lot of the early association studies being picked up very quickly by companies and marketed as a test for propensity to *x* or *y*. As for whether they have any long-term effect, I know some studies done for smoking cessation, which were done out of Oxford, were commercialised by a company to try to indicate which might be the most

effective way of stopping smoking, and whether to use drugs like Zyban or not. They were backed up by good evidence, but I do not think, unfortunately, the commercial service was a success because people did not want to pay £300 to be told how to stop smoking.

Q109 Lord Winston: Is there a place for better public information, do you think, in this area? Again it comes back to this notion of information and dialogue. Is there something which the Department is looking to think about? Presumably, in time, as Lady O'Neill has implied, there is going to be an increasing demand. Lifestyle change may well be a very good thing, but it may not be relevant to what is found on these rather focused and rather selective tests.

Dr Bale: There is certainly a role for public dialogue here. At the moment, we see the Human Genetics Commission taking a lead there. They have been very active in over-the-counter testing. Since 2002 they have been discussing this from time to time, working with some of the regulatory bodies like the Advertising Standards Authority to clamp down on misleading claims that are being made for some services that have been marketed. They are also, I am aware – I am sure they will have touched on this in their evidence – been increasingly interested in the markets that seem to be opening for personalised genomes, of people wanting to have their whole genome sequence scanned. Again, that is a developing area of interest and one that raises a number of issues.

Q110 Lord Winston: That is £1 million at the moment.

Dr Bale: I gather it is coming down.

Professor Davies: It is £300 at the moment.

Dr Bale: The projections are, from studies in things like silicon chips, that it will be a \$10 genome, which is quite staggering.

Q111 Lord Winston: Is anyone doing any monitoring of the Internet? How do we know what should be causes for concern?

Dr Bale: There certainly have been some. A researcher on the Internet tends to go to a well-known search engine and tap in key words. You find a number of services, both coming up on search engines and also being advertised. There is a big difference between those that are being actively marketed in the UK by the UK best companies or their agents. The last time the survey was done was by some academics based at Cambridge and there were very few companies – I think around about half a dozen – marketed actively in the UK. But people can access tests by going over to the States.

Q112 Chairman: The answer to whether there is any systematic monitoring is no.

Dr Bale: No. That is right.

Q113 Baroness Perry of Southwark: I would like to turn to the question of advice on the ethical issues involved. How are the Patient Information Advisory Group and the other advisory groups interacting with the National Research Ethics Service in giving advice to researchers and ethics committees on genetic studies, particularly on issues of consent and confidentiality? This is where I advise you of my interest, in that I chair the research governance group for Addenbrooke's and Cambridge University's clinical school.

Professor Davies: Thank you. Then you will know that we have a National Research Ethics Service, which supports a number of ethics committees who are appointed by the Strategic Health Authorities, including some multi-centre research ethics committees. The National Research Ethics Service and the Patient Information Advisory Group (PIAG) work together quite closely with other partners of the UK Clinical Research Collaboration. They have developed joint guidance for applicants on issues reviewed by both bodies, such as consent and confidentiality, and they are looking at working on further procedural guidance together.

One of the things they have done recently, which is going to be launched before the summer, is the Integrated Research Application System. It is part of the Best Research for Best Health bureaucracy busting, where seven review bodies' applications are brought together and the researcher will put the data into one database. It will do for seven of them, including the National Research Ethics Service, PIAG, the Gene Therapy Advisory Committee and MHRA, so that we are trying to get everyone working together. A lot of the advice is brought together under the umbrella of the UK Clinical Research Collaboration (UKCRC) regulatory governance advice service which we run funded by NIHR with MRC input and advice and support as well, so there is advice there. Of course, under the Health Act going through at the moment, PIAG will be dissolved and its role will move to the National Information and Governance Board (NIGB). The secondary legislation will be laid this autumn.

Q114 Baroness Perry of Southwark: That is very much welcomed and I think is a big step forward in what you call “bureaucracy busting” but the co-ordination of advice on consent and confidentiality does not seem, at least in the experience of the researchers I have worked with, to have gelled yet. There are a lot of sources of different advice.

Professor Davies: They are supposed to have shared views. Many academics have said to me that they find some ethics committees understand the application – Addenbrooke’s being one – rather better than some others, so there is work to be done to educate people.

Q115 Lord Taverne: How many people are employed by this National Research Ethics Service?

Professor Davies: Its budget is £1200 a year. It has a handful of staff in the National Patients Safety Agency centrally and then it supports all the ethics committees around the country, of which there are many. If you would like the detail, I would need to get a note.

Q116 Lord Taverne: I am slightly worried about the huge industry in ethics field which does not always represent good value for money.

Professor Davies: Since I have been in post, we have gone for a single form across all ethics committees, to lighten the load on researchers and be able to move things around. We have an Internet application that is now going to be integrated with the other regulatory people and we have appointed advisers to all the committees to help the chairs move work forwards. We have reduced the bureaucracy and we have also speeded up dramatically the times taken for turnaround from all committees. There are always some that are good, but from all of them. It is a much more smooth service for the recipients of it or the users of it, and I have taken out some costs savings.

Lord Taverne: Good.

Q117 Baroness O'Neill of Bengarve: That is all very welcome news but, in a sense, what drives the complexity is a set of standards, some of them embodied in international documents, which have fanciful notions of the possibility of informed consent. It may not be the notions of consent that are fanciful; it is the notion of the amount of information that can be absorbed by persons, be they patients or research scientists. Is there any way in which you think it is possible to bear down on essentially gestural conceptions of the consent in favour of the bits that matter?

Professor Davies: I find this difficult to answer because I suspect that, as a person, I am exactly where you are: I wonder how often patients do give true informed consent. As a department, I would have to say that we are trying to make sure things are done properly, recognising a pragmatic reality.

Q118 Baroness O'Neill of Bengarve: In which case it is very important not to read the sociological literature that reveals how seldom it is informed.

Professor Davies: I am aware.

Q119 Chairman: So far, the written evidence we have had and the seminar we heard would suggest that, in scientific terms, genomics research is moving at quite a pace. Not only is it moving at quite a pace, but there are implications – Lord Winston referred to the epigenetic aspect, which is crucially important – that this could well transfer into health care, either because of the public getting more knowledgeable in demanding the test once they hear about it or because there are genuine benefits for the population of the test. We then obviously will have some information about the Biobank and other genetic banks. Then there is a gap between that advance and the implication it has on the Health Service and the Health Service organisations to change and be able to deliver and be able to explain, including education. I made the comment that nobody else has given us the evidence about education and genetics being important in what they are doing. Would you agree that there is a gap? Second, I wonder how you felt, when you were writing the evidence to us, about all these different organisations. They may have been necessary at the time when they were set up but are they performing a co-ordinated function? Will they deliver what is required to be delivered in terms of research, translation and health care? Would you have a comment about that, considering you wish to see Britain in the forefront of genomic medicine?

Professor Davies: Let me start with the research and the multiple funders who are being co-ordinated and co-ordinating with each other. I think that works. It looks messy and it is messy, but to have multiple funders, as long as we co-ordinate a certain amount, allows different approaches. Many of us know people who applied to one place and did not like the approach and yet they got the funding from somewhere else who did like that approach. Different bents and approaches are useful. I think the research field, now that we are co-ordinating yet better on things, is fine. You asked about how we will get more pull-through. Of course we have a structure in the NHS now where there has been more and more

devolution: a budget goes out to the commissioners, to the primary care trusts, and to some general practices, to commission the services they want. It is not held centrally and able to be dished out for national programmes in the way that it used to be, and so education and patient demand become more and more important as to how you get things to change. There are two things that need to happen: you need clinicians and a system that is innovative – hence the discussion about Academic Health Sciences Centres. There is not any evidence but we all have a prior belief – and it might turn out to be a Bayesian belief, actually – that they will increase the innovation. Having got innovation and shown that it can work, how do we then roll it out? That is now the role of commissioners. The policy team work with commissioners on selected areas to try to give them some guidance as to what to pick up on.

Dr Bale: That is all I can really say on that. There is a pipeline here and it is at the commissioning end that we need to see professionals demanding this type of service or advocating this type of service with the evidence that has been provided by the scientists and the commissioners. We have not mentioned a group we operate to try to find a forum for this. The Genetics Commissioning Advisory Group is a platform for the laboratories, the professions and the commissioners for the main Strategic Health Authorities to come together to discuss developments in genetics, new service pathways and so on, to try to find a coherence there about how they commission these activities. You also asked about the question around public expectations of genetics and how fast that is moving. Really, from the very earliest days, since we have been actively involved in the Human Genome Project, there has been a huge expectation as to what the completion of the Human Genome Project would give us in terms of information for a health care. Every stage has shown that it is much more complex. For a long time there was no clarity about the number of genes and now we are seeing a completely different way of analysing the other “omics” and epigenetics, as has been mentioned. It is about being quite clear to the public that this is not going to be an overnight

fix; this is a very long-term pathway and programme of investment support and capacity building in the NHS.

Q120 Lord Warner: That answer and everything we have heard today and in our seminar all point to genomic medicine being an additional cost, if not a significant additional cost for the NHS to absorb. That is what it feels like, from the evidence we are hearing. Is there any work going on to identify what genomic medicine might displace, so that we free up some resources to pay for this innovation? At the moment, all we are getting is messages that that is, to put it crudely, “shed loads more money”.

Professor Davies: It depends on the tests you are talking about, does it not? In my field, sickle cell disease, we used to do the genetic tests by looking at the protein, haemoglobin, and it cost five pence per test. As yet, doing it by sequencing, that gene is not cheaper. It will only come in when it becomes cheaper. The added cost is: Is it supplementary or is it alternate? This is an area where the costs of the technology are coming down all the time, and the speed of it, so some of it is likely to add cost but I think you have to look at it case by case and what the advantage is. If you take, as I would in a broad definition of genomics, the treatment of chronic myeloid leukaemia with imatinib, it is a very expensive drug, but it keeps people out of hospital so that they only go in in their terminal phase. You have to think about each example and then look at the cost of putting it in or what it does to the patient journey thereafter and the impact on the service then.

Dr Bale: We have funded, through the White Paper money, the first NHS chair in pharmacogenetics in Liverpool, who has already been doing some work on the pharmacogenetics of certain adverse drug reactions for some of the commonly prescribed drugs. The figures for warfarin and for other drugs prescribed for epilepsy are quite compelling. A very simple test given to a patient before they go on the drug can stabilise the dose much more quickly and with fewer side-effects and hospital admissions. In some cases,

it can even prevent extremely severe side-effects which will be apparent in some populations, in some of the oriental populations, if they have a particular marker they should not have this drug for epilepsy because of the risk of severe side-effects. There is a quite clear cost-benefit there which can be very easily applied at the time as part of the evaluation process.

Q121 Chairman: Thank you very much. If you have any further comments about any of the issues, please could I encourage you to send in more evidence. I do not know if you have any last comments to make.

Professor Davies: I think you have given us a very fair hearing. Thank you.

Chairman: Thank you again for coming here. We appreciate it very much.