HOUSE OF LORDS

Science and Technology Committee

Corrected oral evidence: Innovation in the NHS: personalised medicine and AI

Tuesday 21 April 2026

10.15 am

Watch the meeting

Members present: Lord Mair (The Chair); Lord Booth; Lord Drayson; Lord Duncan of Springbank; Baroness Jones of Whitchurch; Baroness Nicholson of Winterbourne; Lord Patel; Lord Stern of Brentford; Lord Verjee; Lord Willis of Knaresborough; Baroness Willis of Summertown; Lord Winston.

Evidence Session No. 9 Heard in Public Questions 95 - 109

Witness

I: Professor Dame Sue Hill, NHS Genomic Medicine Service.

USE OF THE TRANSCRIPT

1. This is a corrected transcript of evidence taken in public and webcast on www.parliamentlive.tv.

Examination of witness

Professor Dame Sue Hill.

Q95 **The Chair:** Welcome to this morning's Select Committee on Science and Technology. We are continuing our investigation into innovation in the NHS, personalised medicine and AI. In particular, we are interested in the role of genomics and the prospect of developing truly personalised medicine across prevention, diagnosis and treatment. We are very pleased to have as our first witness Professor Dame Sue Hill, who is the chief scientific officer at the NHS and heads the NHS Genomic Medicine Service.

I will start. Dame Sue, you have led the development of the NHS Genomic Medicine Service since it began, building on the work of the 100,000 Genomes project that we heard about from Professor Sir Mark Caulfield. Can you set out for us what the NHS Genomic Medicine Service delivers today and how it benefits patients? What achievements would you highlight? Would you like to start with those questions, please?

Professor Dame Sue Hill: Thank you very much. Just for everyone's information, we have had genomic testing in the National Health Service for a long time. In the 1960s, we were looking at chromosomes down microscopes. Over the period from that time to the start of the 100,000 Genomes project, there had been variable types of genomic testing across the NHS for patients for its application, particularly in rare and inherited disease. What the 100,000 Genomes project taught us was the level of transformation that needed to happen in the National Health Service in terms of clinical leadership, infrastructure and the data and digital arrangements to support whole genome sequencing.

In 2018, when we launched the NHS Genomic Medicine Service, it was building on those years of genetic testing in the National Health Service and the experience of the 100,000 Genomes project. The first thing we did was establish a genomic testing service, which meant that we could offer universal access to genomic testing as prescribed in a national genomic test directory. Previously, access was variable depending on where you were in the country, and, if you overlaid outcomes, outcomes were influenced by access to genomic testing—particularly in cancer.

We also brought cancer genomic testing into our genomic laboratory hub infrastructure and drove some consolidation. The first element was the laboratory infrastructure. The second was the introduction of something we called a genomic medicine service alliance both to drive the embedding of genomics into end-to-end clinical pathways and to lead some research and innovation initiatives, including with industry and academic partners.

We have since built on that model because our seven-year contracts have just come to an end. We have recently procured, through a "most suitable provider" route, seven genomic medicine services, which include the embedding of clinical functions along with laboratory functions and

enablers such as workforce development, data and digital support, leadership of research and service improvement. We have been on a learning journey, and we have evolved the service on the basis of the infrastructure that is required to drive genomics into end-to-end clinical pathways.

Currently, we deliver nearly 900,000 genomic tests in rare diseases and cancer; that equates to just over 200,000 patients with cancer having access to testing. There are 350,000 new diagnoses of cancer every year.[1] We have also introduced what remains a world-leading rapid whole genome sequencing service for acutely unwell children, from neonates through to children aged eight or so. It has a turnaround time of around 10 days, with a diagnostic yield of around 40%. This drives quite challenging clinical decisions in the NHS; for example, when you might introduce palliative care rather than actively treating because of the nature of the mutation that is found.

We have a world-leading foetal exome service, which is driven by the appearance of the developing foetus on the ultrasound taken of pregnant women. It is evolving into a whole genome sequencing service. There is a 30% yield on that, with further developments in both of those two areas to drive a better diagnosis.

In 2021, we introduced, in conjunction with Genomics England, a whole genome sequencing service for the NHS, building on the 100,000 Genomes project. Almost 200,000 whole genome equivalents have been sequenced to date for use in patient care on the NHS; that is continually evolving.

Those are some of the areas I would highlight. We are world-leading in the infrastructure that we have created. I work with many health systems and governments around the world. We are probably around five years ahead of any other country in terms of what is embedded in the National Health Service as a genomic medicine service.

The Chair: Thank you. I know that Lord Patel has a question, but I want to ask one follow-up question. You mentioned access to genomic medicine. We have heard concerns about geographical variations across some parts of England—perhaps across the UK—with some patients being less likely to receive a genomic test that could benefit them. Is that a fair concern? Do you feel that that is being addressed?

Professor Dame Sue Hill: I cannot speak for other countries of the UK, obviously. There are differences in what is offered in genomic testing. In England, we have a national genomic test directory that sets out the national funded offer; that is based on equitable access to everyone who fits the eligibility criteria. This test directory is updated annually on the basis of evidence, which may be generated by researchers working on the UK Biobank data or , by researchers working on the Genomics England database or by the Genomics England diagnostic discovery team

[1] Not all types of cancer require genomic testing.

Associated with the introduction of the Genomic Medicine Service, we established something called patient-level contract monitoring, which means that we collect data on every patient who receives a genomic test. We can relate that data to postcodes, the condition that is being tested for and the testing that they have been offered. This drives both national and regional interventions, especially when there may have been a problem around equity. It has been really important to have that level of data; it has also driven, for example, our projects with the Race and Health Observatory to ensure that we have interventions if access is not at the level we would expect for certain groups in our community.

Q96 **Lord Patel:** I have a question about DNA analysis of tumours as a genetic service. I realise that that is an important service for the managing of cancers, but we cannot meet the target of a 10-day reporting limit for every patient—I understand that only 68% are met, which is way off the target of 98%—without which treatment cannot start. If a treatment is delayed by four weeks, there is a 6% to 8% higher risk of death. In all these discussions, I worry about the hype around what is possible with genomic testing and other testing and the reality of being able to deliver it to the whole of the NHS. We now have huge variations in care; in particular, the people who suffer the most are from deprived areas. How do you manage not being able to meet these targets for all patients?

Professor Dame Sue Hill: Along with clinical experts in cancer across the country, we brought together a consensus workshop to agree on the turnaround times for different cancer types—for example, for blood cancers, separately from solid tumours. In general, apart from those areas—especially in blood cancer, where we need information within three to five days to effect a fairly urgent treatment decision—the turnaround time that was agreed by consensus was 14 days. That has to include the time from when the tissue is acquired, which might be through a biopsy or an operation, through to pathology, with all the pathology assessments they have to do, and then into genomic assessment.

The overall strategy for cancer genomic testing is to provide a comprehensive molecular profile to drive precision medicine decisions at the point of the first treatment, rather than the second or third treatment down the line. We are working closely with pathology colleagues to streamline the pathway because the two elements need to come together. To support that streamlining in NHS England, we have created cellular pathology genomic centres to fast-track the tumour into the other pathology assessments that need to be made, such as immunohistochemistry and other staining for certain types of cancer and then rapidly into genomic testing..

Parts of the NHS are delivering to the agreed turnaround times. Others are not, which is why we have an ongoing service improvement programme to drive the turnaround time down and to look at introducing, for example, circulating tumour DNA so that we can test tumour

fragments in blood. We have introduced this for non-small cell lung cancer—it has driven turnaround times down—and more advanced breast cancer, but there is more to do.

Q97 **Lord Patel:** My next question follows on from what you just said. It is about the 10-year health plan and the shift to prevention, using genomics assessment as part of prevention. The suggestion in both the 10-year plan and the cancer plan appears to be that it is possible, by doing whole genome sequencing, to have a profile of people, using their lifestyle to prevent possible diseases, including cancers, later on. How realistic is that in terms of making precise judgments—apart from, I accept, in inherited genetic diseases where the genes have a clear marker, such as the BRCA gene, as well as in prostate cancer and other cancers? Can you comment on the idea that you can use genomic sequencing and possible lifestyle to predict, at an earlier age, whether someone will develop a disease? How realistic that is for personalised medicine, which the Government want? Although developing genetic medicine holds some of the promises, in your opinion, where are we likely to see the NHS make the best investment for that, and where are there likely to be problems?

Professor Dame Sue Hill: First, from a cancer perspective, the 10-year plan, whether for adult cancers or paediatric ones, focuses on confirmed disease—in the cancer commitment—and on introducing a comprehensive molecular profile that may be from whole genome sequencing.

On the more population health-based approach, currently, as part of the NHS Genomics Population Health Service that we introduced this month, the focus will be on expanding BRCA testing, because we already have high-risk populations whom we are not testing. Our focus will be on the genomic tests where we understand penetrance in populations and the next steps in the care of the patient. The first step will be predisposition genetic testing for cancers. The second will be in other areas such as familial hypercholesterolemia, which has a frequency in the general population, rather than subdividing into different ethnic groups of one in 250, as well as other cardiac and renal tests where we know that there is a need in the population, where we have a well-established and well-understood genetic test, and where there is a pathway for intervention.

The second point, to which I think you referred, relates to polygenic or integrated risk scores. We know that the evidence for their widespread use in the NHS needs to be generated, particularly on the types of public health intervention that would need to be offered—that is, whether people are low, medium or high-risk—and on the infrastructure and implementation that would need to happen at a local level in the National Health Service.

We are taking this forward in two ways: first, agreeing with clinical experts on the high-risk cohorts we want to test; and, secondly, working with Our Future Health and the participants it has recruited and already undertaken genotyping on, in order to start a more evaluative study

based on recruitment from the population at large, rather than targeted cohorts. This is very much in the service evaluation space. Genomics England will, in conjunction with the NHS, undertake an adult population study, where it will probably use whole genome sequencing to understand how that might be applied. In general, through the National Health Service, as it stands at the moment, there are no plans to introduce whole genome sequencing at a population level—only associated with rare disease and cancer.

Lord Patel: That makes sense.

Professor Dame Sue Hill: Yes. As we are investing in testing through whole genome sequencing, where we can also look for additional findings; they might be findings that are not associated with the suspected diagnosis of the patient. In cancer and rare disease patients, we could look for the presence of other conditions—this is the subject of discussion with clinical experts—such as familial hypercholesterolemia. We could also generate from these patients' whole genome sequences a pharmacogenomic profile that contributes to the optimisation of medicines and prevention of adverse drug reactions as part of the population health agenda.

Q98 **Lord Stern of Brentford:** Thank you for that helpful introduction. Can you comment on the role of private testing? Suppose individuals decide, rightly or wrongly—wisely or unwisely—that they want to get a private test. How would the result be used? How would it be helpful, or would it be unhelpful?

Professor Dame Sue Hill: The British Society for Genetic Medicine issued a statement on the use of private testing and bringing such testing into the NHS. In the NHS we follow that professional advice. Often, those private tests have not been performed to the NHS's testing standards. People have opted to have those tests.

There is a second element to this: thinking about the capacity that will be required of the NHS over time. Some private providers—for example, as part of our Jewish BRCA programme—have already been providing some of the testing to NHS standards. That has been fed back to the programme participants and taken place with an agreed set of project aims and objectives, as well as with accountability and deliverability to the standards that are required.

In general, if people have private testing, it usually needs to be repeated on the NHS. That is why the approach from the British Society for Genetic Medicine is that the NHS will not accept the results for use in care. We are increasingly having conversations with the private sector about the role it may or may not play in future.

Lord Stern of Brentford: Would you encourage or discourage private testing?

Professor Dame Sue Hill: Some of the challenges include the fact that—we have evidence of this—the analysis and interpretation of the testing has not been correct. This has led to concerns for individuals, as well as the need for much more extensive testing and analysis on the NHS. At the moment, we would not advocate for that, but we want to work with those industries to see what role they could play in supporting the Genomic Medicine Service in a way that has standardisation, the ability to share data and all the other things we would want to have in place.

The Chair: Lord Willis, who will ask the next question, is online.

Q99 **Lord Willis of Knaresborough:** Good morning. This question follows on from that set of questions. If we are going to depend on the NHS workforce to do more to accept the challenge, does the NHS have enough clinical geneticists, genetic counsellors, bioinformaticians and laboratory scientists to deliver on the Government's ambitions for genomic and personalised medicine, such as those set out in the 10-year plan? What is the scale of the gap, and what are we doing to address it? I want to follow that with another question on nursing, but could you answer this one first?

Professor Dame Sue Hill: Since the 100,000 Genomes project, we have had a genomics education programme, which has been developing education and training interventions for the genomic testing that we have been introducing—particularly to support the ordering and use of genomics by non-clinical genetic specialists, including oncologists, cardiologists and neurologists. It also includes just-in-time resources when a clinician is in the clinic. We have been working with the Academy of Medical Royal Colleges to influence postgraduate medical curricula and the resources that are required. For example, yesterday, I was at the festival of surgery. We were talking about the developments in the genomics education and training interventions by the Royal College of Surgeons. It is an ongoing conversation.

On your specific question, we have around 3,200 people working in the NHS Genomic Medicine Service. We are working with a team that is developing the 10-year workforce plan by modelling the potential gap over the 10 years of the 10-year health plan, along with the commitments set out by the Government in that plan. Part of this will be about moving genomics on from being an exceptionalism and, therefore, having a specialist workforce to undertake all the elements. We will still need genomic scientists, bioinformaticians and others who analyse the data—and those numbers will need to increase—but part of moving genomics on and making it commonplace is developing the broader healthcare workforce, so that they are able to order genomic tests, to understand what to do once a result is available.

This is part of a much bigger plan for developing the whole of the multiprofessional workforce in the use of genomics. Some of that will range from awareness raising right through to much more formalised education and training. The Government's commitment to review all

healthcare professionals' curricula by the end of the decade enables us to embed genomics knowledge and skills into those postgraduate curricula. The area we need to work on more is undergraduate programmes, to ensure that we can develop a workforce that is already genomically aware—even though, because it is moving at pace, ongoing upskilling will be needed. That also needs to be coupled with the developments in data and analytical skills as we bring all this together with the genomic information

Lord Willis of Knaresborough: What are the top requests that you have made to the Government to ensure that we can recruit, train and retain the skilled workforce we need? You may do not remember this but, 10 or 12 years ago, when you and I last met, you told me off for not demanding enough of graduate nurses to look at the future. You were absolutely right. Nurses are never mentioned, as if they do not exist, but surely they are crucial for working in the skilled workforce to deliver this to patients.

Professor Dame Sue Hill: I remember our conversation all those years ago; thank you for reminding me of it. Nurses and midwives are critical in our being able to deliver the genomics commitments in the NHS. That is right across the care continuum, from tertiary and secondary care into primary and community care—and, increasingly, in terms of their role in neighbourhood teams. We have a programme in nursing and midwifery that is being led by a chief nurse, and we have been developing education and training programmes for that staff group for some time, but this will not stop. We will have to do more, particularly as we expect that it will not be limited to nurses. We expect, for example, pharmacists and others to be genomic champions in the community. It is an ongoing piece of work focusing on the multiprofessional workforce, but it is about recognising that data and analytical skills will also need to be part of that.

The Chair: Baroness Willis is also coming in online with the next question.

Q100 **Baroness Willis of Summertown:** You have partly answered this question. I want to ask about training the next generation of medics, medical students and undergraduates. I am aware, both from personal experience and from being in a university setting, that we are looking at a six-year window to train a medic. Where in that medical training should they start on advanced genomics, if we are really serious about having the next generation understanding and being able not only to do the experiments but to interpret the results appropriately?

Professor Dame Sue Hill: First, we need to start early and influence the Medical Schools Council to include genomics in the undergraduate curriculum. At that stage, some of it will need to be not just straightforward genetics and genomics but applied genomics, so that students understand how it will be utilised in future. Secondly, we have already had fourth-year and fifth-year medical students getting experience by working in the Genomic Medicine Service; we need to formalise that better for those who are interested. Then our challenge is

to do a review of the postgraduate curricula so that—this goes for all healthcare professionals—a continuum of education, skills, development and competence is developed from that early undergraduate level.

Baroness Willis of Summertown: Can I push you a bit further on that? You said, "For those who are interested", but should not every single medical student have an understanding of this? It is a bit like AI. This is the future of so much of what we are doing. I am concerned that only those who seek it will get it.

Professor Dame Sue Hill: My apologies—what I meant was that every medical student in the curricula needs to be genomically aware, as well as aware of data, AI and, for example, how wearables will be part of the data sources that medics may have in future. On fourth-year and fifth-year medical students, we want to have an offer that means that they could come and spend some time with the Genomic Medicine Service if they wanted to choose that as an option for their elective. We need to make that more systematic.

The other point I want to make is that we are working with industry because we often train bioinformaticians in the NHS, then industry recruits them. We are working with t industry to look at similarities in education and training requirements, as well as how we could train together so that everyone's needs are met. This recruitment of bioinformatics staff from the NHSoccurs because we have a very structured scientific training programme for bioinformaticians in the NHS.

Baroness Willis of Summertown: Broadening that out slightly beyond the medical students, what about clinicians and the people who are managing all the other aspects of it? Do you think that we need more training outside of these specialist genetic services?

Professor Dame Sue Hill: Yes. On making genomics commonplace, you will be aware of the ambition in the 10-year health plan that, by 2035, 50% of all healthcare interventions will be genomically informed, together with data, AI and other analytics. This means that there is a huge education and training task for us to do. Part of that work will be to determine what is required by whom, because not everyone will need to be an expert in genetics; for example, going forward our nursing and midwifery staff will need to have some of the skills of our genomic counsellors. Much the same has happened in CBT—cognitive behavioural therapy—where other members of the delivery team have developed some of these skills.

The future will involve developing the workforce in mainstream specialties, as well as primary and community care, so there is a huge education, training and workforce development piece. Part of what we will do is develop a training needs assessment as certain types of genomic testing and their application are rolled out within the NHS. Part of the work will be what Lord Patel asked me about: the rollout of polygenic risk scores and the use of that type of predictive risk assessment. Even if we

roll things out into the NHS in an initial a service evaluation way to start with, we will develop education and training interventions to support that rollout.

Baroness Willis of Summertown: I have one more tiny question. Who would deliver that? Would it be the Department for Education or the National Health Service? Where does the DfE come into this?

Professor Dame Sue Hill: Some of it will be through the education and training that is eventually commissioned and contracted by the Department of Health and Social Care as NHS England moves into it. [2]Oth er elements will be through the Department for Education and the programmes.[3] The third element will be through the in-service training in the NHS that is delivered as for example part of the postgraduate training in medicine, , which is why we are working closely with all the medical royal colleges, including the Royal College of General Practitioners, specifically to develop the education and training interventions that are appropriate for primary care. We already have a master's in genomic medicine, which we introduced during the 100,000 Genomes project and which is delivered by higher education institutes in England.

My discussions with academic health science centres in England indicate their willingness and enthusiasm to work together to introduce the required education and training programmes in genomics in their colleges and institutions. That will be how we will take this forward. It will be multi-pronged, but it will require everybody with an interest in education and training to be involved.

Q101 **Lord Drayson:** Our inquiry is using genomic medicine and AI as a tool to understand why the NHS struggles to adopt proven innovations across the service. In your evidence, you have already mentioned how the genome-sequencing service is the world-leading infrastructure five years ahead of the competition. However, as you discussed with Lord Patel, this is not the universal picture across the UK. Can you help us understand the systemic barriers that lead the NHS to struggle to adopt proven innovations that have been shown, in islands of excellence within the NHS, to make a real difference to outcomes but do not get adopted across the NHS as a whole?

Professor Dame Sue Hill: I would like, if I may, to use genomic medicine and what we have been doing to illustrate some of the elements that we have tried to put in place to drive the adoption of innovation.

You will be aware that an innovation ecosystem review was undertaken by NHS England. The Genomic Medicine Service was highlighted as an exemplar of good practice, partly because of the mechanisms we have to drive innovation in the NHS. Some of those are key components of having a system that is ready to adopt innovation. We have been clear about what we want to test for and how we will deliver it. For example,

[2] This includes the resources that are available from the genomics education programme.
[3] That is to say, those delivered by higher education institutions and other organisations.

as we move forward, having set out our priorities for innovation in genomics, we will work with a health innovation network to act as a front door for some of the companies that want to develop genomic interventions.

There have been a number of different developments, but they do not fit with the model of delivery—that is, being able both to deliver in a cost-effective way for the NHS at scale and to provide equitable access across the NHS in England. To achieve success the first thing is being very clear about the purposes and the priorities. The second thing is having clinical leadership, at the Genomic Medicine Service, we have created directors of innovation to work more locally and across the geography. We have seven genomic medicine services that cover populations of between 5 million in the south-west to 11 million in our central and southern infrastructure.

Those directors of innovation also work with interested companies across their patches to help develop the evidence for commissioning in the NHS. Finally is having access to resources and linked to that, we have established genomic networks of excellence that are bringing together NHS England funding with the funding that has already been put into the system by research funders—for example NIHR, MRC, Wellcome and others—together with industry partners, to increase the potential resources that are available. They look at the areas of unmet need and generate the innovation solution. Importantly, they also provide the health economic assessment to help in the decision making on commissioning . These networks of excellence work in two-year windows. We have just completed our first two years. Many of those innovations are now ready for adoption in the National Health Service within that two-year window.

A key feature of the Genomic Medicine Service is that it is nationally commissioned. We have a national genomic test directory, which covers any innovation or evidence. For example, just over 18 months ago, there was evidence of the discovery of a new gene associated with neurodevelopmental disorders, with one publication in May and one in July. By November, we had that testing available on the directory so within less than 6 months the evidence/innovation was adopted.

Lord Drayson: In the description you just gave of how you have addressed some of the issues—it was really helpful, so thank you—you hinted about the problems you are overcoming. I would like to probe a couple of your comments a little deeper. You said that you have found challenges, in that the innovations do not fit the model of delivery on the clinical pathway. This suggests that different clinical pathways are being adopted in different parts of the NHS, so coming up with a solution that can be scaled economically is challenging. Is that what you are getting at?

Professor Dame Sue Hill: Not particularly. What I am saying is that, if we have to deliver a genomic testing service within our given budget,

what we want to do with that budget is deliver testing as efficiently and productively as we can—that is, at the lowest possible cost base and with the highest possible quality associated with it.

Some of the solutions that come to the table and come across my desk are often small-scale innovations that cost a lot of money to implement in the NHS and in an end-to-end clinical pathway. Therefore, part of the conversation that we are having is with the Association of British HealthTech Industries, for example, is about setting out what we are particularly looking for, whether that is scalable, being able to deliver at a low price point or being able to test for multiple targets.

Lord Drayson: I want to make sure that I understand this. When you say that it is on a small scale, is it the case that the solution just cannot cope with the numbers you require?

Professor Dame Sue Hill: Yes, that is right.

Lord Drayson: Therefore, because it cannot cope with the numbers, it cannot be done cost effectively, so it is a non-starter.

Professor Dame Sue Hill: Yes. It also requires modifications to the clinical pathway to introduce some of the testing. Therefore, we are trying to define the priorities and the conditions for adoption in the NHS, including the evidence that is required to get a particular test into the service or on to the NHS genomic test directory.

Lord Drayson: We have heard evidence from other witnesses in our inquiry describing a risk aversion in the workforce towards innovation. It is a culture where there is a punishment for failure but nothing for the adoption of innovation. Professor Sir Mark Caulfield highlighted the fact that the 100,000 Genomes project achieved effective adoption because the NHS was involved from the research stage. Can you comment on that? Do we have a cultural problem with innovation in the NHS?

Professor Dame Sue Hill: As the chief scientific officer, I head the profession of 58,000 healthcare scientists. I can say that those scientists adopt innovation all the time. Sometimes, what prevents the adoption is the funding to support the adoption locally. That is one of the challenges. They do not fall short in coming forward with ideas for the adoption of innovation, or even developing the innovations themselves for adoption.

One thing that was brought out in the innovation ecosystem review—this is why I made that point earlier—was alignment with the commissioning system. Often, the evidence is generated from research studies, but it is not always generated in a way that will influence a commissioning decision around how it changes a pathway, makes a pathway more cost-effective or makes the outcome for a patient better, as well as on whether it is safer or whether there is a different way of using it within a pathway. That evidence is often not provided, which is why we work closely for example with Genomics England on its newborn sequencing generation study to understand how that would be adopted should it get

through the UK National Screening Committee.As well as influencing the impact assessments and evaluations to ensure that there is evidence for adoption in the National Health Service. , , We have been working right from the start with Our Future Health—you will talk to Raghib Ali shortly—to ensure that, when there is an agreement to feed back the result to participants to influence their care inthe NHS, any testing by Our Future Health will be done to NHS standards so that it can be assessed for adoption in the NHS.

Lord Drayson: If I have understood you correctly, you are saying that the system does not enable individuals to have the headroom to be as innovative as they could be, rather than there being a fundamental culture of risk aversion.

Professor Dame Sue Hill: It depends on where you are. In genomics, we do not have a shortage of people finding the headroom. That is why we created the directors of innovation: to give headroom to look at innovation. Leadership in innovation is a key part of how you drive adoption. You will probably be aware of innovation adoption specialists in hospitals in America, where they drive the adoption of a whole raft of different technologies in some of the bigger healthcare systems in the US.

The Chair: Lord Duncan, do you want to come in briefly?

Q102 **Lord Duncan of Springbank:** Very briefly. I am struggling a little, as what you describe is an extraordinary possibility. However, the cost of doing that seems very high. Whenever we read about the NHS, we hear of an NHS in crisis, where there is not enough money and where the challenges are on the front line. Presumably, the hope for your area is that you will save money, ultimately, but you will almost certainly not save money in the early part. Therefore, the early part of the journey will require significant cost investment before any savings come. Is there a risk of you ending up not being able to complete the journey because you have high costs but you do not then have high savings at the end?

Professor Dame Sue Hill: You make an important point. This is why, in having a national budget for the Genomic Medicine Service, we can continually review the available budget, in a way other parts of the service cannot. , We have an annual update of the test directory. We can remove a test that is no longer required because we have replaced it with a higher-level genomic test that gives us the same or more information, rather doing six different tests that might mean the cost is greater. We can understand the cost per test , because of the level of contract monitoring that we have in each of the seven areas, but we are about to introduce a national tariff price for genomic testing; that will be under continual review because the driver of that price is about making the delivery efficient and cost-effective .

With the ongoing rapid developments in genomic technologies tThis will require a case to be made for ongoing investment to make them available to the eligible population but, importantly, we have to couple

that with the ability to describe the benefits to the system—both the health economic benefits and the broader benefits, which reside not just with health but with social care and, sometimes, with education and disability. Therefore, part of the work that we need to do going forward is about understanding the much broader benefits of genomic testing—especially doing so earlier, when you can reduce with detection the amount of disability that people experience.

The 100,000 Genomes project told us about the cost of a diagnostic odyssey in the NHS, so part of this is being able to describe that because it will mean savings in other parts of the services that wrap around genomics. It is about being able to describe that in a way that makes sense to the investment system for both capital and revenue.

Q103 **Baroness Jones of Whitchurch:** How much of this is due to the deeply structural challenge that we have in the NHS? We have trusts, a complicated commissioning landscape and so on. To what extent are those things holding innovation back? As we know, the NHS is a huge organisation. You have described a complicated process for establishing research priorities. Meanwhile, in the UK, we have such a good reputation for research and innovation.

There is such a lot of work going on out there. It somehow feels as though those people who are being innovative are finding it really difficult to break into the NHS structure. They might get taken up by one trust but then have to start again with another trust down the road. Are the confident that, with the structures we have, we can give the innovators on whom we want to capitalise—that is a huge opportunity for us in future—the opportunities that they deserve, because they are doing some fantastic work out there?

Professor Dame Sue Hill: From a genomic perspective, because it is a nationally commissioned service and the budget is held nationally, we have the ability to adopt innovation and do so, as I said, very quickly—within a year, even, if we need to do so and the evidence suggests that we should adopt it. That might be, for example, to support with genomic testing a new NICE-approved medicine.

There are new developments—I am not leading on them, but I am aware of them—associated with both the 10-year health plan and the life sciences sector plan, such as NIHR's innovation catalyst, to drive a pathway and generation of evidence for adoption into the National Health Service. Also, as part of its refresh, the work of the Health Innovation Network will help generate the evidence for commissioning. The evidence for the commissioning, as well as evidence on where it makes a change in existing pathways or outcomes for patients, is the information that we actually need.

Baroness Jones of Whitchurch: How does an innovator in the private sector, the university sector or wherever break into that and say, "Look, this is the work we're doing, and we really think that it will be transformative"? How do they influence your decision-making?

Professor Dame Sue Hill: As I said earlier, from a genomics perspective, rather than the broader perspective of the NHS, we set this out—the areas of unmet need where we need to work with innovators, including academics and industry partners. Those are set out as priority areas. For example, we are about to introduce a network of excellence in using a particular type of sequencing in brain cancers.

Baroness Jones of Whitchurch: You have explained the genomic side of it quite well, as well as the fact that that is nationally commissioned, so I feels like that is sorted. It is all the other things that are going on, in terms of innovation in the NHS, about which I am slightly more concerned.

Professor Dame Sue Hill: Planned developments from the NIHR, from the work of the Health Innovation Network and in the commitments that were set out in the 10-year health plan will help bring clarity on the type of evidence that needs to be generated for integrated care boards, for example. This is not my area to speak about specifically. I am sure that others in the system—whether it is the NIHR or the people who are responsible for the Health Innovation Network—can set out what they are doing. Part of this is about ensuring that there is the right evidence to support the commissioning system.

In a separate part of my world, in my chief scientific officer role, we do exactly that. We do calls with industry for areas we are working on, such as antimicrobial resistance t diagnostics. We then work with different partners or with the industry to generate evidence for the different innovations. Then we produce the guidance for the system, in terms of adopting that technology—that is, for the operating system in the NHS. We have been doing the same in blood group genotyping, where we are trying to look at expanded blood groups to match blood better.

Q104 **Baroness Nicholson of Winterbourne:** Might it be possible for you to launch a novelty programme, as it were, to see patients—they are, after all, the largest cohort and block of people inside the NHS system—in an innovative programme as a sort of pilot scheme? You do not have just the patient: you have their families, their friends and all the other people around them. In that way, you might transform the NHS's approach to innovation, provided that it was built into that and there was a successful privacy outcome for those points that needed to be kept private.

Professor Dame Sue Hill: Yes. There is no doubt that patients and their families have a very good understanding of the innovations they would like to see. That is not necessarily the case in all of medtech; it is across the board in innovation in healthcare. It illustrates the importance of having patient and public involvement, which is exactly what we do at the Genomic Medicine Service at every level of the system. They influence the innovative approaches that are being looked at across our seven areas, which range from education and training innovation right through to, for example, the acceptability of a new innovation for diagnosing patients with rare disease.

Baroness Nicholson of Winterbourne: I was thinking that you could work out a small, public pilot programme. If it worked, it could be spread right across the different elements of the NHS, which, as we have already said, are very diverse at the moment. That is just a thought.

The Chair: We are going to ask some questions about AI now.

Q105 **Lord Drayson:** Our inquiry is focusing on AI, in part because of the speed at which the science in AI is moving and the potential change it can bring to health outcomes but also because, fortunately, the UK is still a world leader—it is in the top three—in AI science. Therefore, this area has the potential to be a real driver of economic growth. In your role as chief scientific officer, what is your view on the role that AI should be playing in the NHS? Can you give us some understanding of how AI systems are procured and how the commissioning is done? You have mentioned the success in genomics, in that there is a national commissioning system, which has been key to driving adoption. What is the picture in AI?

Professor Dame Sue Hill: Looking specifically at the evidence for the utilisation of AI in genomics—we are testing different solutions in the NHS at the moment through our networks of excellence, as is Genomics England—and if there were a decision that it would contribute to the work of the Genomic Medicine Service on finding unmet need or analysing genomic data, for example, we would be able to commission that nationally as part of the Genomic Medicine Service because the commissioning is national. As for the arrangements on the commissioning of AI in the NHS in general, I would have to get back to you with a response on that, because how that would eventually be commissioned does not sit with me.

Lord Drayson: Okay. Building on Baroness Jones's questions about the balkanisation—if I can use that term—of procurement in the NHS, do you think that there is a case for removing the ability of individual trusts to procure and centralise NHS procurement for certain areas of products, systems and software, such as electronic patient record systems?

Professor Dame Sue Hill: Again, I am not deflecting in any way, but answering that question is not really within my remit. What I can say is that, in the context of genomics, we have to drive data quality up. To use AI, we will have to drive up our ability to collect data to the right standard. Then we will have to work on removing some of the silos where data is kept. If we want to bring together genomics, other diagnostics and other outcome data, that will require those silos to be broken down. From a genomics perspective, we are working with EPR providers, such as Epic, to ensure that the standards to which genomic data is collected is at an international level to support both data sharing and the use of AI more generally, because we know that it requires high-quality data.

We have the UK Biobank, Our Future Health and Genomics England in this space, all of which will be testing some of these technologies. Separately, the question of whether it is value-based procurement that

has been introduced as a result of the 10-year health plan commitment, or other types of commissioning arrangement, is probably something that somebody else would need to answer.

Lord Drayson: May I push you a little further? In your excellent evidence today, you have mentioned examples of how the infrastructure has been set up. You just mentioned Epic as an example of a software company. Like the majority of software companies used by the NHS, it is supplied largely by US-dominant software companies. You have talked about how the procurement process aims to get value for money but can you give us some sense of the potential for the adoption of new data science and artificial intelligence, as this wave of innovation is hitting the NHS? What consideration is there in the procurement process of the relative merits of buying software from one of these enormous, very rich companies from the United States and from UK-based developing businesses?

Professor Dame Sue Hill: Can I give you the example of working with Epic and Cerner, which is what we will be doing?

Lord Drayson: It is now owned by Oracle.

Professor Dame Sue Hill: That is right. Even in this country, different NHS organisations—this is around the genomic element of Epic—had to enter into separate arrangements with Epic each time they wanted to introduce some of the genomic data standards and other elements. What we are trying to do globally, working with an organisation called the Global Alliance for Genomics and Health, is to work with both Epic and Cerner to introduce international data standards into those EPR systems as part of a global discussion towards implementation. This will prevent individual organisations having to do it and the individual costs associated with that. It is for a very particular innovation, but it is working towards a very particular application in the genomics field. There is an appetite from Epic, for example, to consider this and to introduce those particular changes. EPR systems themselves specifically are not within my remit, though.

The Chair: We understand that. Lord Booth is coming in online.

Q106 **Lord Booth:** I am not with you in London today because, last Wednesday, my husband had CAR T-cell therapy. One of the major concerns we have heard is that personalised medical treatments such as CAR T-cell therapy can be very expensive, especially when they are in the early stages of innovation. My question for you is: are NICE's cost-effective frameworks and the NHS's commissioning models fit for purpose in supporting innovation in personalised medicine? Do they need to change? Additionally, are there regulatory barriers from the MHRA that need to be addressed?

Professor Dame Sue Hill: Thank you for that question. You may or may not be aware that there is now a taskforce working on rare and emerging therapies. It is looking at all the elements you have outlined there,

including the regulatory elements; it is working with MHRA on streamlining those processes, as well as with NICE on the evidence that is required for its value assessment.

This will be increasingly important in the area of precision medicine, especially because of the developments in the manufacture of some of these agents, not just in this country but elsewhere, for N-of-1 therapies. There is an active set of discussions that are ongoing. For this committee, further evidence could be provided on the work of that group and what it is looking at. That would partly addresses your question, and we are looking at how flexible the approach to regulation and NICE processes needs to be and how things need to change to respond to new and emerging therapies. Equally, from a genomics and other diagnostic perspective, how do we ensure that patients are stratified as part of their routine care, rather than having to do that separately and delaying their access to these precision medicines if they are part of clinical trials ?

Lord Booth: In his evidence to us, Professor Caulfield explained that pharmacogenomics—using genetic information to guide which drugs are prescribed—is one development in personalised medicine that could be deployed quickly in the NHS. We know that it has been scaled up, but why is it not yet routine in the NHS? What would you do to get us there?

Professor Dame Sue Hill: It is routine for four indications. Currently, we perform around 40,000 pharmacogenomics tests every year on the NHS; those are associated with the use of some drugs in the treatment of cardiovascular disease, the use of some chemotherapy agents and the use of antibiotics, because a particular type of antibiotic can cause deafness. It is routinely available and funded for all eligible patients on the NHS.

There are several lists of drugs that could be tested for adverse reactions on the basis of genomic information . The challenge is around which ones would give the best outcomes for patients in the NHS now, in terms of reducing adverse drug reactions and preventing, for example, hospitalisation or multiple visits to general practice. We have done a primary care study in pharmacogenomics looking at commonly prescribed medicines, such as some anti-depressant medications, statins. We have shown that there can be modifications in the prescribing of those drugs for around 30% of patients, and we are beginning the process of rolling that out to the NHS at large through the NHS genomic population health-based service.

We are also looking specifically at applications in mental health, where patients often receive polypharmacy—multiple medications—with, sometimes, severe adverse drug reactions. There is evidence that, if we performed some pharmacogenomic testing in those patients, those adverse drug reactions could be reduced. We are doing this with a very careful, case-based approach, in terms of the evidence and the potential benefits for patients, by looking at what infrastructure is required to be put in place to deliver this at scale. For example, if we wanted to test all

the NHS patients who took a medicine called clopidogrel, we would have to prepare for around 1.2 million tests to be performed.

Although we know that pharmacogenomic testing will make up a large part of the 10 year health plan 2035 ambition, we are working on the infrastructure that will be required to do this and on how we can introduce it in a systematic way, so that the system is ready to receive it and those people who need to manage those patients can manage them and act on the results. The digital and data developments also mean us having to work with primary care systems. We have at the moment a mechanism that creates a data store and pushes data when it is asked for, but we will have to develop the primary care systems to ensure that they can access that data.

The Chair: I am conscious of the time. You have answered so many of our questions. I will ask my colleagues to be as brief as they can so that you are not kept too long. Lord Winston is online with the next question.

Q107 **Lord Winston:** Thank you very much, Dame Sue, for your extremely interesting evidence so far. One of the rather more eye-catching considerations announced by the Government is the possibility of genomically screening babies at birth for various risks. Can you comment on that and give me your own views on whether that is a wise, or even possible, thing to deliver?

Professor Dame Sue Hill: The generation study is being led by Genomics England in partnership with the NHS. You may be aware that they are looking for just over 200 conditions. To enable that to happen, 200 clinical pathways have needed to be established, because these babies are often not treated at birth but when they present to the system sometime later.

A major amount of work has been done, including with NICE, to ensure that there has been agreement on the interventions, if they are treatment interventions, as well as on some of the confirmatory diagnostics. To date, 138 "condition suspected" results have been seen in the study, and 58,000 participants have been recruited. Where this will head is, first, in trying to find rare disease early—there is no doubt that disease is being found early—and, secondly, in understanding what needs to be put in place in the NHS to ensure that those babies can be cared for in the system and followed longitudinally as part of their care.

Part of the work of the generation study is generating the evidence for whether it would meet UK National Screening Committee approval. Until we understand the outcome of that—and of the ongoing work that is happening to understand the follow-up studies that may be needed if for example further evidence is required—it is very difficult to comment on whether it is something that would be adopted over time.

Lord Winston: That was a bit of a cautious answer, if you do not mind my observing so. What about informed consent? What is the issue regarding counselling people who are being screened? You said, "Start

early", which I think is right, but we could start with the embryo. We could start early with women who have had a stillbirth or other problems as well.

Of course, one of the issues will undoubtedly be that, although you may have 150 mutations in which you are interested at the moment, all of us carry fatal mutations in our genome that are not likely to be expressed unless we marry the right partner. That will be a huge problem. One of the problems will be the effect on teenage mental health—that is, finding out that you had been tested for a gene that might cause a disease but probably never will be expressed?

Professor Dame Sue Hill: That is why there was a very careful approach to the conditions that were selected for use in the study. Those are genes that are highly penetrant and associated with the development of the disease. You are right that it is linked to confirmatory diagnostics for the condition. At the moment, it is not going into areas where there may be uncertainty around the level of expression. It is still early days for this study. As I said earlier, we have a foetal exome service that is sequencing foetal DNA on the basis that there were clear abnormalities on an ultrasoundAs we look at this whole area, we will need to consider what else will be done on foetuses, as well as what might be done at birth, in terms of what that pathway looks like.

We are, for example, working on stillbirths and providing genomics in that perinatal mortality area, but the evidence is still being generated.. However, you will have also seen and be aware of some of the emerging evidence of treating in utero, which means that we also have to look at the testing that needs to support the use of these treatments and ensure that it is appropriate, can be performed and is acceptable.

Lord Winston: Let us take one area where it has been really effective. Thirty-five years ago, the first babies were born—they are now still alive—after screening for a specific disease that had killed an earlier sibling. That was a good example of innovation in the NHS.

Professor Dame Sue Hill: Yes.

Lord Winston: Of course, as you have rightly pointed out, commissioning has been really difficult for patients who carry those defects; it is still not accepted, I think, even by NICE. It has been pretty difficult to get that through. You have not commented yet on NICE. Can you do that?

Professor Dame Sue Hill: I can comment on NICE from a genomic perspective. We have an NHS England/NICE framework so that we are both clear on what the horizon looks like for the precision medicines that are likely to come through.[4] Over the past 18 months, we have had to introduce genomic testing for 12 precision medicines, and those numbers are increasing all the time. One aspect is horizon scanning. Secondly, it is

[4] For example, health technology assessment (HTA) and other NICE programmes.

about trying to agree—especially if it is in the areas of genomics, precision medicine area or pharmacogenomics—what some of the priority areas should be, then discussing up front the implementation considerations that they will need to take into account.

For some medicines, we have a framework for how we price the cost of genomic testing for NICE purposes . In the NHS, we are providing testing at a greater scale and for multiple diseases because it is more cost-effective to do that rather than single testing for a single outcome or a precision medicine.

Q108 **Lord Patel:** I think that my question, which relates to the national cancer plan and genomics, has been answered, so I will not go back to it. However, I would like your comment on something you said in answer to Lord Booth's question about personalised medicine. You talked a lot about using genomics for personalised identification and the treatment of diseases, particularly cancers. You also talked about pharmacogenetics to identify individuals for whom certain drugs may not be appropriate and referred to polypharmacy, which is a big issue.

Of course, it would be right to personalise medicine that does the individual the greatest good. However, it is expensive as opposed to the standard practice of giving treatment to the majority, which does the majority, but not everyone, good. One aspect is expense against personalised medicine. The other is in polypharmacy, for instance. A good example is an intelligent pharmacist in a hospital, who will look at all the medication prescribed to an individual and say, "Remove that and that because they interact with each other", so they remove the side effects. What are we chasing here?

Professor Dame Sue Hill: On your last point, I was relating polypharmacy to severe mental illness, where those patients can get severe adverse reactions.—

Lord Patel: I mean in all diseases.

Professor Dame Sue Hill: I do not think that, at the moment, the evidence is there for moving into polypharmacy and pharmacogenomic testing for everyone. However, for certain groups that get quite severe adverse drug reactions, there is evidence for moving into that area. That is why my response was, in terms of where we go with pharmacogenomics, we could do everything, but it has to be very carefully considered in terms of the cost benefits and the outcomes that can be achieved.

On your comment about precision medicines, what we know from the evidence is that, for cancer, accessing a precision medicine early on in the patient pathway leads to a better outcome.

Lord Patel: Yes, but that is based specifically on identifying the genetic mutation against which a drug would be effective or not prescribing a drug that is not likely to be effective if the mutation is not there. I

understand that. Trying to do personalised or precision medicine for everybody through more and more testing, whether that is for pharmacogenetics or genomes as usual, is possible but very expensive.

Professor Dame Sue Hill: Yes. That is why I think that it will have to be on the basis of the evidence for the greatest benefits. I have made the point several times about the importance of health economics in this.

The Chair: Our final question comes from Lord Verjee.

Q109 **Lord Verjee:** Thank you very much, Dame Sue, for a very thorough evidence session. We have a vast wealth of experience in the NHS, helping to integrate innovative genomic medicine into routine care. Our committee will ultimately produce a report that makes recommendations to the Government. What would be your top recommendations to ensure that the NHS can integrate innovations such as personalised medicine and AI? What does the NHS most urgently need to make this a success?

The Chair: That is a difficult question to answer, but what would be your top three recommendations that you would like us to make?

Professor Dame Sue Hill: I will start as I began: in the area of genomic medicine. We have a strong genomic ecosystem in this country. We have a strong NHS Genomic Medicine Service and robust infrastructure. We have world-class cohort studies through Our Future Health and our UK Biobank, as well as other studies and the resource that has been created through Genomics England and the National Genomic Research Library.

So we have a very strong base on which to ensure that we remain world-leading. Part of my request would be that we continue to remain world-leading in this area. We must understand the key elements of maintaining that world-leading position, which needs to be supported through a range of different initiatives and agreement on resourcing. That would be part of my response.

We should also continue to adopt cutting-edge technologies, not just in genomics but across diagnostics, and to look at the potential for some of these technologies to remove some of the other testing that we do in the NHS so that it remains an effective service that gives the best possible outcomes for patients.

Part of the challenge with personalised medicine is ensuring that the data sources can join up. For AI, we must have data that is collected to the right standards, which is no longer siloed and which we can join up, along the patient pathway, in order to link the diagnostic with the personalised treatment and intervention so that we can collect the outcome. Looking at that whole pathway will mean us being world-leading in personalised medicine. Other countries focus on the front end—the diagnostic—but not on how it links to either the treatment interventions or the outcomes, what it means, and the outcomes for people and their family members.

The Chair: You have answered many of our questions. This session has been extremely informative, so we are very grateful to you. We will now

pause and prepare for the next part of the session. Thank you very much indeed.