

The 100,000 Genomes Project: transforming the NHS

Meeting minutes

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHSA

Parliamentary attendees

Jo Churchill MP
Dr Philippa Whitford MP
Dr Sarah Wollaston MP
Baroness Browning
Baroness Emerton
Baroness Finlay
Lord Freyberg
Baroness Howe
Baroness Masham
Lord Sharkey
Lord Taverne
Lord Warner
Baroness Watkins

External attendees

Barrett, Jeffrey	<i>Wellcome Trust Sanger Institute</i>
Barrett, Michael	<i>Judge Business School, University of Cambridge</i>
Bitner-Glindzicz, Maria	<i>Institute of Child Health & Great Ormond Street Hospitals</i>
Blackburn, Laura	<i>PHG Foundation</i>
Bloom, Theodora	<i>BMJ</i>
Bowers, Sarion	<i>Wellcome Trust Sanger Institute</i>
Brice, Philippa	<i>PHG Foundation</i>
Brooking, Stephanie	<i>Oxford Nanopore Technologies</i>
Burton, Hilary	<i>PHG Foundation</i>
Carragher, Fiona	<i>National Health Service England</i>
Caulfield, Mark	<i>Genomics England</i>
Compton, Richard	<i>Independent</i>
Davie, Edward	<i>Royal College of Pathologists</i>
Fleming, Amy	<i>Association of Medical Research Charities APPG on Medical Research</i>
Fowler, Tom	<i>Genomics England</i>
Fromen, Peter	<i>Illumina Inc.</i>
Gortana, Stefano	<i>PHG Foundation</i>
Kroese, Mark	<i>PHG Foundation</i>
Lishman, Suzy Royal	<i>Royal College of Pathologists</i>



Personalised Medicine

All-Party Parliamentary Group

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHS

Livingston, Andrew

McDougall, Zoe

Nevin-Ridley, Katrina

Parry, Vivienne

Perrin, Nicola

Searle, Beverly

Sharpe, Rowena

Sherley-Price, Edward

Turnbull, Clare

Vance, Jennifer

Wells, Tom

Wilde, Simon

Wilson, Julia

Shield, James

Yadi, Hakim

Roche Diagnostics

Oxford Nanopore Technologies

Genomics England

Genomics England

Wellcome Trust

Unique

Cancer Research UK

Genomics England

Genomics England

PHG Foundation

Department of Business, Innovation and Skills

Genomics England

Wellcome Trust Sanger Institute

Macmillan Cancer Support and APPG on Cancer

Northern Health Science Alliance

Minutes

Welcome and overview - Jo Churchill MP

- Welcome to all attendees
- Stressed her commitment, and the commitment of the APPG, to the value of genomic medicine and its crucial role in the future of the NHS
- For patients with rare diseases and cancers, the data collected via the 100,000 Genomes Project could help lead to earlier diagnoses and improved treatments
- Genome sequencing has the potential to benefit many other areas of medicine as well and has already proved important for detection of hospital infections and antimicrobial resistance
- The challenge will be bringing this technology and understanding to mainstream hospitals
- There is no guarantee of success of the 100,000 Genome Project but it is the challenge and opportunity that we must seize and it is right and proper that the government shows itself willing to invest

Introduction to the 100,000 Genomes Project - Prof Mark Caulfield

- While focused on the English NHS, it has been confirmed that Scotland, Northern Ireland and Wales are joining this programme with funding from their respective devolved nations
- We are sequencing as much as we can read of the genetic code; focusing on rare inherited diseases, cancers and infectious disease
- Infrastructure has grown to include the first genomic medicine sequencing centre and a central data centre
- Rare diseases affect about 3 million of the UK population - working on over 200 diseases, nominated by NHS, researchers and industry, and we will begin to return several thousand diagnoses over the next few weeks



Personalised Medicine

All-Party Parliamentary Group

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHS

- We are working on many of the common cancers across the whole range. More specifically, we're looking at optimizing how we can use small pieces of cancer tissue to get genomic diagnoses back to patients that will allow us to choose the best treatments for them
- We're also working on infection - we have already sequenced the genome of 3000 bacteria that cause tuberculosis. This understanding may help us benefit from the next generation of anti-microbial medicines.
- Working with countries around the world, including Canada, France and Australia
- We have formed a consortium of companies to help shape Project data to fully deliver this project

Genome sequencing: the clinical perspective - *Prof Maria Bitner-Glindzicz*

- Commitment as a genomic medicine centre is enormous: in North Thames, we've committed to providing 15,000 samples from patients during this Project, over and above the routine clinical work we do every day
- Working across seven trusts, we have been asked to provide up to 5 tubes of blood per patients with multiple replicas, meaning hundreds of thousands of extra samples on top of the clinical laboratory medicine done daily
- We're doing very well thanks to the committed clinical staff
- We have a number of recruiters in our centre, we work with other research teams to increase recruitment and we host weekly late-night recruitment clinics as well as Saturday morning clinics
- Staff are fully committed to the programme and are extremely dedicated to their patients suffering with rare diseases. They believe in the benefits of the 100,000 Genomes Project and patients know they have the support they need
- There is still great unmet need out there and the 100,000 Genomes Project is certainly worth investing the time and effort in from the point of view of families and NHS services

Genome sequencing: the patient perspective – *Edward Sherley-Price*

- After a difficult birth, a range of tests and years of uncertainty, the journey towards a diagnosis was frustrating and emotionally challenging
- Through the Deciphering Developmental Delay, the prequel of Genomics England, Alysia was given a diagnosis
- It was a real milestone for understanding and an emotional relief that Alysia's condition was not the result of any particular action or inaction and that Alysia's sister was not vulnerable to the genetic condition
- The diagnosis allowed for more effective medical intervention and the provision of the appropriate medicines to address Alysia's particular condition
- The diagnosis also gave a surge in hope: that future medicines could be available, that future developments in genetic testing may help others and that awareness and understanding may improve the lives of those with genetic conditions. This hope has really driven the family forward

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHSA

Question and Answer - Jo Churchill MP

Baroness Browning questioned how Genomics England interacts with NICE

Prof Caulfield responded by highlighting the persistent discussions held between Genomics England and the regulators, including NICE. He added that the university research engine has been aligned with the health system to streamline innovation and translation, in addition to including key champions of the process. He also stressed the belief that Genomics England would have the necessary evidence to satisfy NICE or other regulators

Sarah Wollaston MP questioned what Genomics England is doing to ensure data security is maintained while facilitating the crucial process of data sharing

Prof Caulfield stressed the importance of maintaining public trust in stored data. Everyone in the programme operates under informed consent; patients are told how the data will be stored and used. Patients are motivated to share by their experience of worry and fear about their genetic conditions. Affected patients are the strongest proponents of data sharing

Prof Bitner-Glindzicz added that clinicians explain the reasons why medical researchers, biotech and pharmaceutical companies need patient data; vast majority of patients are more than happy to share. Many have children and really want their data to be shared and the opportunity to help other families

Edward Sherley-Price confirmed that data security is very key and the participant panel rigorously tests Genomics England on that. The results from inquiries reiterate the high-level of security and willingness to share data

Jo Churchill MP also commented on how important it is for patients to fully understand why their data needs to be shared and what it actually achieves. This is only the start of the journey

Dr Suzy Lishman asked how to ensure there are the right number of pathologists on the ground with the right training to help deliver the benefits for patients

Prof Caulfield said that, within the context of a wider investigation by NHSE into what will happen after the 100,000 Genomes Project, they are looking at combining a number of budgets (with molecular pathology concerns at the core of that thinking); pathology services have changed such a lot and are needed most right now

Lord Warner added that there has been some consensus around the need to consolidate services, without which the investment and capabilities to take advantage of many of the things Prof Caulfield and Bitner-Glindzicz have talked about will be undermined. There are benefits to consolidating services in fewer sites to get more volume out of the resulting capabilities

Baroness Masham highlighted the problem of late diagnosis and the consequences of extra expense and stress on families, as well as patchy services across the country. The question focused on access to the 100,000 Genomes Project across the country

Prof Caulfield stressed the importance of the 13 genomic medicine centres which provide greater geographic access to their work

Prof Bitner-Glindzicz noted that families can often be quite separated in their geography, with multiple members living in different towns. More recently, this has been effectively addressed and participation has since been widened



**Personalised
Medicine**
All-Party Parliamentary Group

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHSA

Edward Sherley-Price added that the participant panel continues to grow and the continual marketing and driving of the centres to propel awareness and recruitment is important

Baroness Finlay questioned whether there had been a needs assessment regarding the size of the backlog of potential patients and what is being done to drive awareness of who to refer, when to refer and the preparation necessary prior to referral to avoid unrealistic expectations

Prof Caulfield highlighted a category of 'ultra-rare unidentified disease' that exists for unique cases. Furthermore, an All Wales genomic medicine service will help boost access and awareness. Charities have increasingly been incorporated into the awareness raising process. Regarding, needs assessment, we know for many diseases about half the people do not achieve diagnoses (about 1.5 million people) but we will eventually get diagnoses for many of these people (25-30% new diagnoses from the whole genome in rare disease which will then be broadened out)

Hakim Yadi questioned to what extent UK expertise in this field are being exported internationally

Prof Caulfield emphasised that the UK trade industry is already working on this as is Genomics England and NHSE. For example, a collaborative with British Columbia offers unique opportunities. Collaboration overseas is very important for access to patients

Lord Freyberg asked why out of 10,000 biopsies only 6,700 reach the UK Biobank

Prof Caulfield explained DNA sample requirements and sometimes there just isn't enough in a sample. Genomics England are working with Illumina to bring down the sample size necessary for testing to help improve these results. Furthermore, the traditional preservative used in the process also damages the tissue, reducing quality. Work is being done on this and a new care pathway is aimed at delivering fresh tissue

Clare Turnbull stressed the many technical challenges of collecting and testing samples

Baroness Watkins questioned when attention will turn to mental health

Prof Caulfield explained there are certain rare diseases associated with mental conditions that are being addressed but the cost of the technology for more common diseases is difficult to justify. Efforts are being made to bring the price down so these programs can be used more widely. The architecture of these diseases are also markedly more complex and will take extensive work

Julia Wilson asked how detailed diagnoses will be married up with new drug development

Prof Caulfield highlighted the clinical interpretation partnership that has resulted in 2400 scientists and clinicians from around the world working to make sense of all the data. A section of the data centre will also be dedicated to moving from data to drug targets. Genomics England has also developed various genomics consortia that enable access to extra info to facilitate outcomes

Hilary Burton questioned what programmes are in place to evaluate clinical achievements

Prof Caulfield pointed to the clinical interpretation partnership that includes a range of evaluation programmes



**Personalised
Medicine**
All-Party Parliamentary Group

Chair:

Jo Churchill, MP

Co-Chair:

The Rt. Hon Lord
Norman Warner

Vice Chairs:

The Rt. Hon. Lord
Philip Hunt
Lord Narendra Patel
Chi Onwurah, MP
Sir David Amess, MP

Treasurer:

Lord George Willis

Secretariat

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN

Partners

Sanger Institute
The BMJ
NHS

Theo Bloom questioned what is being done to ensure GPs know what to do with information flowing from the 100,000 Genomes Project

Prof Bitner-Glindzicz admitted that scaling up a workforce is tricky. There are a number of Masters course to encourage clinicians to learn more about genomics. There has also been investment in education of GPs and clinicians. The Project itself has driven interest in the subject

Hilary Burton wondered how these programmes will work with 130,000 specialists in addition to 230,000 GPs in the country who need to understand genomics. The Clinical Champions programme for example helps raise awareness but more detailed educational work is still lacking and is needed

Jo Churchill stressed the important role of medical journals and other organisations to educate clinicians and raise awareness

Michael Barrett questioned how the UK is learning from industry partners

James Shield wondered what to expect in cancer care in the next five years

Jo Churchill asked Edward to identify key patient outcomes he would like to see

Prof Caulfield said that industry partners are looking at early genomes, helping shape the data centre and in talks about potential projects to do alongside Genomics England. In cancer, there will be better precision medicine and a better understanding of how to treat and when

Clare Turnbull emphasised that the difficulty of consent is a persistent barrier, that pathology needs more attention, that data should be held in one centre location and that drug availability is still a concern

Edward Sherley-Price asked for a joined-up approach across departments and regular communication regarding advances in medicine

Jo Churchill closed the meeting with a commitment to the challenge as well as the inherent opportunity of the 100,000 Genomes Project

phgfoundation
making science work for health



BMJ

