



Public Health
England

The UK 100,000 Genome Project

Dr Philip Monk



Genomics England (GeL)

About the 100K Genome Project

We are a new company set up by the Department of Health to help deliver the 100k Genome Project first [announced by the Prime Minister](#) David Cameron in December 2012.

This project will sequence the personal DNA code – known as a genome – of up to 100,000 patients over the next five years. This unrivalled knowledge will help doctors' understanding, leading to better and earlier diagnosis and personalised care. Based on expert scientific advice, we will start by tackling cancer, rare diseases and infectious diseases.

The company will manage contracts for sequencing, data linkage and analysis, and set standards for patient consent.

"The UK will become the first ever country to introduce this technology in its mainstream health system."

Genomics England was announced by Jeremy Hunt, Secretary of State for Health, as part of the NHS 65th birthday celebrations on 5 July 2013.

He said: "The NHS has a long track record as a leader in medical science advances and it must continue to push the boundaries by unlocking the power of DNA data.

"The UK will become the first ever country to introduce this technology in its mainstream health system – leading the global race for better tests, better drugs and above all better, more personalised care to save lives.

"Genomics England will provide the investment and leadership needed to dramatically increase the use of this technology and drive down costs."



Prime Minister,
David Cameron



Secretary of State for Health,
Rt Hon Jeremy Hunt MP



Genomics England - Mission

- 100,000 patients with rare inherited disease, common cancers and pathogens from the NHS in England
- Whole Genome Sequencing
- Generate health and wealth
- Legacy of infrastructure, human capacity and capability
- World-leaders in Genomic Medicine
- £100m funding over the next 5 years



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Proposed pilots

Pilot 1-2000 Rare Inherited Disease WGS

30x depth - 2014

Pilot 2- Lung Cancer from NHS

1000 somatic (50x) and 1000 germline (30x)

Informatics, annotation and alignment pilots

Pilot 3 - Pathogens

Led by PHE

TB and Hepatitis C



What will GeL offer for pathogens?

Scientific validity – commentary on the proposed pilot and scientific advice

Support to achieve the best possible costs for sequencing

Support for scientific grant applications



Genomics England – who are the people?

Officers: Sir John Chisholm (Executive Chair)

Mark Caulfield (Chief Scientist), Nick Maltby (Company Secretary), Jim Davies (Informatics), Louise Hiller-Holmes (PA)

Board: Professor Dame Sally Davies CMO, Kevin Dean (Cisco), Professor Sir John Bell and Vivienne Parry,

Advisory Committees:

Science: Sir John Bell, Bioinformatics: Kevin Dean and
Ethics: Mike Parker



What are the steps and timescales?

Building on the national MIRU / VNTR genotyping service and latterly the successful work at Oxford University led by Professor Derrick Crook and using the Illumina platform MiSeq sequencers purchased by PHE

We will be carrying out sequencing in 3 NHS laboratories

Brighton

Oxford

Leeds

And one PHE Regional Mycobacteriology Laboratory

Birmingham



How does the sequencing system work?

When the isolate flags in MGIT, DNA is extracted and sequenced

The sequence reads are transferred via the cloud to Oxford

The reads are assembled in the Oxford pipeline

A report is generated and sent from Oxford to the referring laboratory for onward transmission to the clinician

The report details

Species, Drug resistance prediction and relatedness to other specimens held in the database for public health purposes



What else

- Sequence the isolates from all MIRU / VNTR clusters from the last three years
- Sequence back catalogues from the centres in the pilot
- Sequence all of the MDR isolates



Clinical phenotype

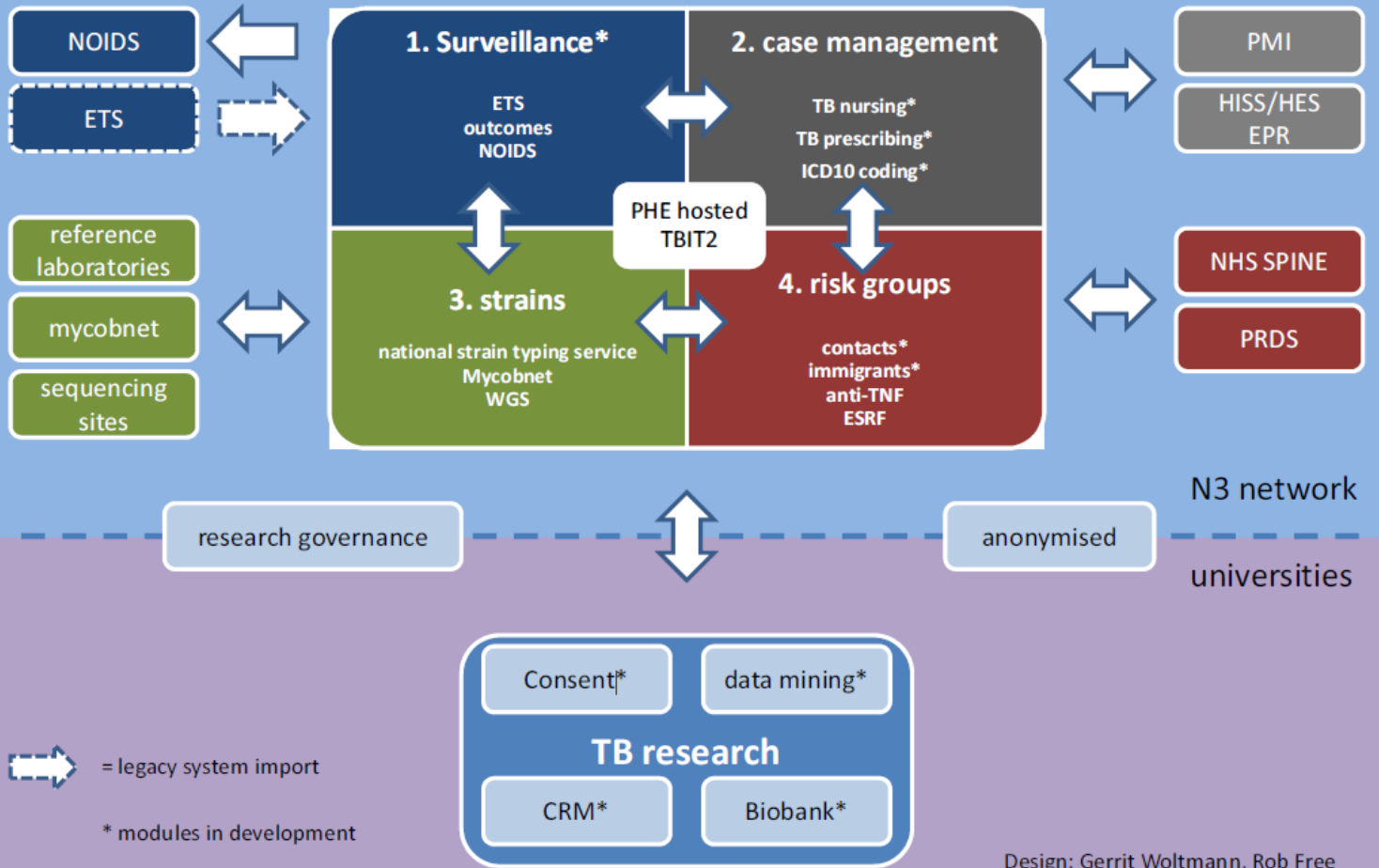
- Establish automated links to a clinical management system

This will allow

- Surveillance - links to national surveillance system
- Clinical management
- Research access



Integrated TB IT model for the UK





Conclusion from recent NEJM editorial

We need to work together to ensure that research progresses, that regulatory policies are developed, that patients' rights and needs are addressed, and that clinical use of genomic information is based on rigorous evidence.