



Monday 18 September 2017



views & analysis

Genome project puts England at cutting edge of precision medicine – and ethics debate



Andrew
McConaghie

January 21, 2016

A bold initiative aimed at making the NHS a world leader in 'precision medicine', England's 100K Genomes Project is advancing rapidly. It is already helping the first patients – but Genomics England knows it must convince the wider public of its value, and retain trust on data security and medical ethics.

Andrew McConaghie

The government is currently locked in an angry dispute with junior doctors in England about plans to create a 7-day National Health Service (NHS). Meanwhile more and more hospitals are falling into financial deficit, with chief executives warning that the system could collapse thanks to an unprecedented funding squeeze.

It is against this background of a health service in crisis that the bold and ambitious 100K Genome Project was launched last year. Its aim is to make the NHS a world leader in using genomic research and insights to improve patient care.

This optimistic vision of the future clearly jars when contrasted with genuine fears for the health service in the here and now, as well as deep-seated public doubts about the Conservative government's stewardship of the NHS.

However the project could genuinely help the NHS maintain its relevance in the 21st century. Its critics frequently portray it as an unreformable relic of a past age, and a system no longer fit for purpose. The 100K Genome Project is an attempt to get the health service to be a leader in 21st century healthcare, to help it focus on preventing certain diseases by understanding their genetic factors. This could be welcomed by most British taxpayers, who consistently rank the NHS as one of the country's most highly-prized institutions.

Nevertheless, because it is pioneering, the 100K Genomes Project carries considerable risks – such as a leak or hacking of confidential patient data, or perceptions that it is being exploited for commercial purposes.

However no-one is any doubt that the future of medicine will one day be based on genomic data – the vision is that the NHS, its patients and the UK's science base can all gain by embracing it.

Genomic medicine

The goal is to sequence 100,000 complete sets of DNA by the end of 2017. This will create an unparalleled database of genomic data, helping researchers understand the genetic basis of diseases better, and accelerating progress towards 'precision medicine'.

The sequencing of the whole genome of individuals began last year, using blood samples provided by volunteers from around the country. The organisation leading the project, Genomics England, chose patients with rare diseases to be its first priority, and has quickly moved on to providing diagnosis and treatment tailored to the first patients given a full genetic profile.

The government has announced today that the first cancer patients will now be recruited to the project.

"Genomics is the future of medicine and the sequencing of cancer DNA confirms why the UK is a global leader in this field," said Secretary of State for Health Jeremy Hunt.

"Over half a billion pounds has been invested in genomics to ensure that NHS patients continue to benefit from the prospect of better diagnosis and better treatments."

A pharma research consortium

Genomics England has even organised a huge two-day Festival of Genomics conference in London this week to help promote the project and its benefits.

The initiative is also aimed at boosting the country's leading position in medical research – but the government is sensitive to any suggestion of commercial exploitation of the DNA database.

It has made clear that data from the project will be retained by Genomics England, and not sold on to commercial researchers such as pharma or biotech companies. But of course the project needs the expertise of industry researchers to unlock the potential of the data, so its solution has been to invite companies to create a research consortium called the GENE Consortium.

Ten companies (AbbVie, Alexion, AstraZeneca [AZ], Biogen, Dimension Therapeutics, GlaxoSmithKline, Helomics, Roche, Takeda, UCB) signed up to the consortium in March 2015 and will spend a year scoping out the 'most effective and secure' way for industry to use the data.

Pharma research and genome company leaders will get the chance to speaking alongside NHS leaders and academic researchers at the

Festival of Genomics today. This includes AZ's Mene Pangalos, head of its innovative medicines and early development biotech unit, as well as executives from Illumina and Seven Bridges Genomics.

Another key partner in terms of medical research is charity Cancer Research UK.

The charity's chief executive Sir Harpal Kumar says the comprehensive mapping of patients' DNA will allow new ways to prevent, diagnose and treat cancer more effectively to be developed in the future.

Sir Harpal mentions new targeted treatments such as Roche's skin cancer drug Zelboraf (vemurafenib) as an example, as it targets the BRAF V600 mutation present in some patients.

"We hope that, one day, treatments based on the specific characteristics of the tumour, including the genetic make-up, can be offered to all patients – and this latest phase of the 100,000 Genomes Project is another important step towards this."

IT infrastructure – a risky business

Such a groundbreaking project clearly requires a major IT and data infrastructure, and the government has signed deals with a range of expert firms; in gene sequencing (Illumina) data management (LabKey Software) and clinical interpretation bioinformatics (WuXi NextCode), among others.

However the UK government has repeatedly performed very badly in procuring and delivering such large-scale IT projects, and will be under scrutiny again.

The project must overcome many technical barriers, from collecting a high-quality DNA sample to its sequencing, secure data storage and then use in computer systems which help doctors interpret and act on the data.

Also hugely significant are data security and consent issues. The government has been caught out once by a public and medic backlash against care.data, its plan to create digital health records. The row centres on whether or not NHS England gave patients sufficient information about the database (and the opportunity to opt out) before its launch, as well as concerns about plans to make the data available to academic and pharma industry researchers.

While this row remains unresolved, Genomics England has pressed ahead with the 100K Genomes Project, and has so far sequenced the entire genomes of 6,000 people.

Illumina is an acknowledged leader in genome sequencing, and is overseeing the huge task of sequencing the 100,000 genomes. All the data, both clinical and genetic, will be deposited in a secure national data centre. Access to identifiable information will be limited to treating clinicians, while access to anonymised data will require prior ethical approval. Nevertheless, the risk of a hacking attack or a leak

of data is impossible to rule out, as demonstrated by recent high-profile cases.

Will it pay off?

Launched in 2012 with over £300 million investment, a further £250 million in funding for genomics was allocated in late 2015 as part of Chancellor George Osborne's spending review.

This signals that the initiative is seen as a flagship project, particularly when Osborne is deepening austerity measures in public spending beyond healthcare.

These sums remain relatively small compared to the £100 billion-or-so spent every year on the NHS in England in total, but it must, all the same, avoid looking like a vanity project.

In the long term, there is no guarantee that investment in genomic medicine will help the NHS save money. It's far too early to predict how genomic medicine will evolve. By helping to diagnose conditions far earlier, it should allow preventative medicine to be practised. But it could just as easily increase overall costs by uncovering many conditions that currently go untreated.

A long-term commitment

Finally, Genomics England is also aware that the project has to be able to demonstrate direct benefits to patients as quickly as possible. It says that, unlike much genetic research, the 100K Genomes Project allows patients to be told about "clinically actionable" mutations once they have been checked and validated.

The project recorded its first successes in December, when children at London's Great Ormond Street Hospital received life-changing diagnoses thanks to its genomic profiling. However further information about the children's conditions was withheld, clearly signalling government caution about divulging genetic medical details.

The country's Chief Medical Officer, Dame Sally Davies, also plans to focus her annual report on genomics when it is published towards the end of 2016. This will apparently aim to provoke public debate about genomics, and "build more public understanding and trust" in the project. This will be essential, as the UK will only become a leader in this revolutionary field of medicine through sustained investment.

About the author:

Andrew McConaghie is pharmaphorum's managing editor, feature media.

Contact Andrew at andrew@pharmaphorum.com and follow him on [Twitter](#).

