Can we be the world leader in genomics? Anna Scott looks at where we are, where we need to be, and how we can get there.

"The age of precision medicine is now and the NHS must act fast to keep its place at the forefront of global science," England's Chief Medical Officer Professor Dame Sally Davies has written in her annual report *Generation Genome*, published in July this year to wide acclaim.

In future, all NHS patients should be able to access whole genome screening and expect it just as they expect an MRI scan today – everyone with cancer or a rare disease should be able to get faster, better treatment – the report states.

The IBMS is even launching two new molecular pathology qualifications this year, which are intended to directly support the vision for personalised diagnosis and healthcare offered in the report.

Despite the UK's status as a world leader in genomic medicine, its full potential has yet to be realised, Professor Davies says. Her report makes seven recommendations that will enable genomics to be made mainstream in all elements of the NHS, from wards to screening policy, including establishing a centralised and national network of genomic laboratories, a ministerial-chaired National Genomics Board, and genomics training offered to all clinicians.

The current situation

In England, over 30,000 people affected by rare and inherited diseases, cancer and infections have already had their whole genome sequenced as part of the 100,000 Genomes Project, run by Genomics England. NHS patients can also receive

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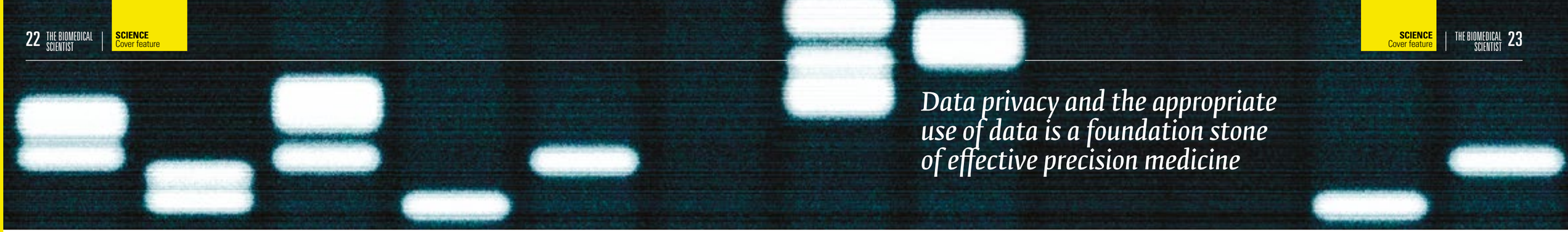


Data privacy and the appropriate use of data is a foundation stone of effective precision medicine

genetic testing at 25 regional laboratories in England.

"We've already begun to set up the infrastructure, education and training and systems within the NHS that are needed for routine commissioned genomic medicine," says the project's Chief Scientist, Professor Mark Caulfield.

"Genomics has the potential to transform the way we care for patients – moving away from a 'one-size-fits-all' model to personalised medicine based on an individual's molecular basis of health and disease."

We've already seen this kind of tailored approach to healthcare with single gene tests for lung, breast and colorectal cancers to inform treatments that are tailored to their genetic makeup, according to Cancer Research UK's senior policy manager, Emlyn Samuel.

"Some patients with lung cancer have a specific mutation in their *ALK* gene and the NICE-approved drug crizotinib – already available on the NHS – targets those specific cells," he says.

The end goal

"There is huge potential here, not just in informing treatment and care, but in informing research as well – to better understand diseases, leading to new treatments," Emlyn adds.

But making genomics mainstream is no longer simply about sequencing 100,000 human genomes linked to quality human data and enabling greater learning about disease, says Professor Caulfield. "The big picture is in building a unique research database, thus enabling a powerful learning system able to provide better outcomes for patients. This requires system-wide standardised data and consolidation of clinical and diagnostic data for each patient."

The UK needs to be able to have NHS genomic testing and precision medicine on a consistent basis and those capabilities need to be delivered in a timely manner, "if we are going to see a transformational impact", according to Mike Standing, Global Life Sciences and Health Care industry leader for Europe, Middle East and Africa at Deloitte.

"The general public should also have a clear understanding of the value of and challenges around genomics and precision medicine and there must be consensus on issues of privacy," he adds.

100,000 GENOMES PROJECT

- ✔ The project will sequence 100,000 genomes from around 70,000 people. Participants are NHS patients with a rare disease, plus their families, and patients with cancer.
- ✔ The aim is to create a new genomic service for the NHS – transforming the way people are cared for. Patients may be offered a diagnosis where there wasn't one before.
- ✔ Combining genomic sequence data with medical records is a ground-breaking resource. Researchers will study how best to use genomics in healthcare and how best to interpret the data to help patients.

Reaching the goal

Sequencing the first human genome, completed in 2003, took 13 years and a global effort. "The genomics sector today requires the same level of collaboration to analyse and interpret data and uncover genomic associations to disease," says Professor Caulfield.

The sector also needs collaboration between patients, healthcare professionals and academic research, says Mike, so that improved genetic testing and precision medicine are delivered at the same time as current services are maintained, and new jobs and globally-competitive companies can be created in "what will be a very important sector".

Genomics England was set up as a private company to run the 100,000 Genomes Project by the government in 2012. "At the very start, we recognised the need to share knowledge and learning across all sectors – academia, industry, government and the NHS," says Professor Caulfield.

It then established GENE Consortium – a pre-competitive industry collaboration aimed at aligning the needs of the commercial sector with the project, ultimately ensuring that genomic discoveries are translated into medical treatments as quickly as possible.

"GENE has generated a wealth of understanding around how all sectors can collaborate more efficiently, driving the growth of genomics and helping to deliver more effective care. Now that the consortium has ended, we have established a collaborative forum to learn from and build on its legacy," adds Professor Caulfield.

Pharmaceutical multinational AbbVie has been involved. "These efforts will enable new medical research to provide a better understanding of the underpinnings of human disease and hopefully lead us to tomorrow's breakthrough medicines," a spokesperson says. "Public and private partnerships are critical to making this a reality."

Collaboration

It's also critical that a patient's perspective is considered, Mike says. "We need to construct this collaboration so that it safeguards the patients, and patient privacy, but it also uses the collective skills of these different organisations to build effective and secure precision medicine.

"Data privacy and the appropriate use of data is a foundation stone of effective precision medicine, and unless that is preserved, trust will be lost and the whole process will slow down. The public has to have confidence in the system and it has to be led by the public sector," he adds.

Genomics England is part of the Global Alliance for Genomics and Health (GA4GH), a collaboration working to set data-sharing standards and tools. Approved researchers from academia and industry can access de-identified data in a secure environment.

"This demonstrates how it's possible to maintain NHS patient data confidentiality while ensuring that we maximise the opportunities to improve understanding of the causes and treatments of disease through genomic research," Professor Caulfield says.

"Without the involvement of industry and their research, the NHS would not be able to get the new medicines, treatments and diagnostics for patients that should come from the 100,000 Genomes Project."

The pharmaceutical sector already develops tools, such as a molecular diagnostic test and companion tests, to go with their drugs, according to Emlyn.

"The commercial sector is driving technological advances, which is driving lower costs, making it more cost-effective for the NHS to purchase these types of machines and technology."

The cost

But if providing DNA testing as standard across cancer care, for example, would lead to early diagnosis, this could result in greater expense for the NHS – already struggling to cope under reduced budgets. While whole genomic sequencing of an individual currently costs about £1,000.

"We know that the cost has come down substantially and it is becoming more affordable," says Emlyn. "But there should be a spectrum of treatment. We can test whole genomes all the way down to single genes."

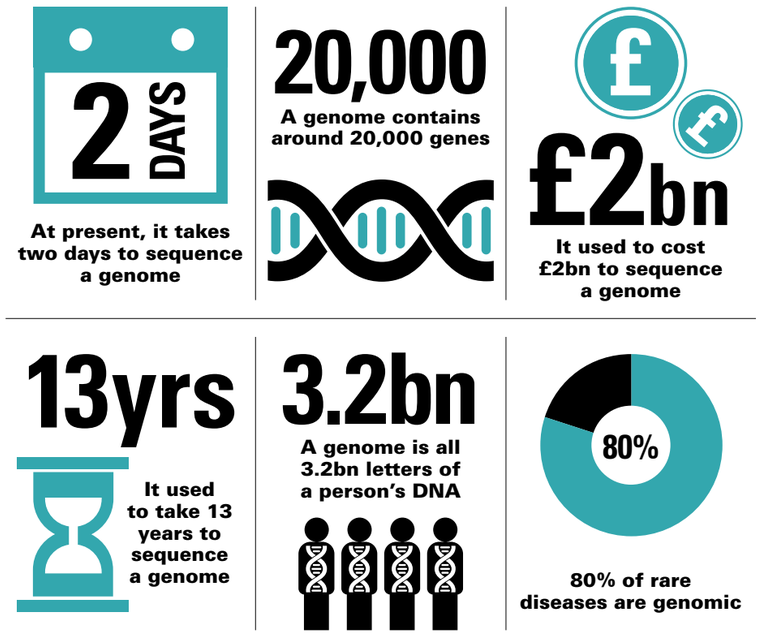
To put this into context, Mike says, all healthcare systems are under significant constraints at the moment – it's not uniquely the NHS. Nevertheless, innovation remains crucial.

"The success of National Institute for Health Research has really shown the ability of the NHS to drive innovation," he adds. "And the NHS' support of the Accelerate Access Review recommendations (specifically designed around how to transform the delivery of precision medicine to patients) is something we can be optimistic about."

The government has taken a proactive view of understanding the importance of innovation in precision medicine and actively thinking across organisations about how the country can "move this forward and build and maintain a leadership position globally", Mike says.

That genomics is being taken seriously by government is crucial for it to be embedded in the NHS, to inform patient care and improve outcomes. BMS

GENOMICS IN NUMBERS



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