Straight talk with…
Sir John Chisholm

Last month marked the sixty-fifth anniversary of the UK National Health Service (NHS), but the British healthcare system is nowhere near retiring. In fact, the publicly funded organization is rejuvenating its role with a new push for genomic medicine.

On 5 July, the UK government announced the launch of a new nonprofit company set up by the Department of Health called Genomics England, which aims to sequence 100,000 whole genomes, with linked clinical data, from people with cancer, rare diseases and infectious diseases—all by the end of 2017. Leading the effort is executive chair Sir John Chisholm, an engineer turned research administrator who until last year served as chairman of the Medical Research Council (MRC), the largest of the UK's three life sciences–focused research councils. **Elie Dolgin** spoke with Sir John about how the NHS plans to roll out its ambitious genetic check-up.

Do you think the timeline of 100,000 genomes by 2017 is doable?

Yes. We have a very ambitious project, something that we believe is a first in the world. And if we succeed with it, it will be something of huge value, not just in the UK, but to medical science in general.

What sequencing platform will be used?

We haven't chosen that yet. We are looking at all the sequencing companies on the market at the moment, but we have yet to publish exactly the structure in which we will be asking for bids. We will be doing that soon. In the first instance, we will be running one or more pilots and then we will be going on to the full program.

Besides sequencing, what will Genomics England do?

Our project also encompasses the capture of clinical data. So, we will establish a clinical data infrastructure and an access point so that our users can tap into the data set in a secure and safe environment with fully anonymized data in order to run the whole genomes against the clinical data provided.

You talk about the data being anonymized, but there have been a number of studies published recently showing that genetic sequences stripped of their identifying information can still be traced back to individual donors. How do you plan to deal with this privacy issue?

One of the key things we're doing is we will be using the very best and very latest anonymizing techniques. But the other important thing is that our data will all sit behind a National Health Service firewall. We will not be passing the data to anyone to take away and mine it for purposes other than that for which is was intended. Our users will bring their algorithms and their processing techniques into our side of the firewall, and they will take away only their processed results. So, we will have a pretty good understanding of what's going in within our safe area.

One of those users could be a pharmaceutical company, as a series of freedom-of-information disclosures revealed earlier this year. That makes some people uncomfortable. What do you say to critics who worry about the commercialization of personal medical information and genetic data?

This is an entirely voluntary, opt-in process. If patients feel uncomfortable about that in any way, they needn't opt in, and the treatment they'll get in the hospital or clinic they're in will be no worse for that.

Prime Minister David Cameron in December 2012 pledged £100 million ($150 million) for the effort that is now Genomics England. That works out to around £1,000 per genome, never mind all the ancillary costs like the ones associated with the clinical infrastructure, which is a pretty tight budget. Do you expect the costs of technology to come down, or are you looking for additional funding elsewhere?

Both. There are people who are extremely interested in what we're doing who would like to play in this program, plus the technology is moving on and we are watching that closely. We're hopeful that for the bulk of our activities we will intercept the technology at a point that is more affordable than it is now.

The UK already has another ambitious genetics-related project in the UK Biobank, which has collected tissue samples from half a million Britons, and those samples are now being genotyped. Besides the technological differences—microarrays versus whole genomes—do you think these projects might be redundant?

No, they're different projects. The one you're referring to is a longitudinal project, and our project is one with a higher focus on disease states—the patients are already in the clinic with very specific symptoms. Having said that, we will then follow them through their own progression.

How do you think your time at the MRC prepared you for this venture?

In the time that I was at the MRC, one of our huge opportunities was to translate the outstanding scientific achievements that poured forth from many laboratories in the United Kingdom into clinical and pharmaceutical products that became available to the population at large. That 'translation agenda' was a huge part of my tenure, and this project [Genomics England] is a natural extension of that.