News story: Genome sequencing project reaches the halfway mark

The 100,000 Genomes Project was launched in 2012 and has now reached the halfway mark – sequencing 50,000 human genomes from 40,000 patients.

Patients have already benefited from their participation in the project:

- participants with rare diseases have received faster diagnoses
- cancer patients have received personalised treatment programmes only made possible by the project

The project aims to provide better insight into the cause of diseases and how diseases develop in each individual. This will ensure that medicine is more targeted and there will be fewer unwanted side effects.

While many developed countries are working on genomic medicine initiatives, no other has the reach and impact of the 100,000 Genomes Project. Patients are recruited through care and treated through routine channels thanks to the unique structure of the NHS.

Of the 50,000 genomes mapped so far:

- 8,000 are for cancer patients
- 42,000 are for rare disease patients and family members

Breast, brain, colorectal, lung, prostate and renal are among those cancers to have been successfully sequenced.

Whole genome sequencing gives a more complete picture of the precise genetic changes causing an individual's cancer. It opens up a greater range of treatment options.

Early analysis has found genetic changes in more than 60% of cancer patients, which could potentially provide new therapies through clinical trials for some of these patients.

Health and Social Care Secretary Jeremy Hunt said:

> This incredible achievement shows once again why the UK is a world leader in genomic medicine.
>
> We're backing our world-leading scientists and clinicians in the NHS to push the boundaries of modern science and embrace new technology – using data to transform the lives of patients and families through quicker diagnoses and personalised treatments.
>
> It is testimony to the hard work of the clinicians and scientists across the NHS and volunteers for the project that we can continue

> to harness the very best of the NHS and remain at the forefront of this pioneering field.

Sir John Chisholm, Executive Chairman, Genomics England said:

> The 100,000 Genomes Project was a stunningly ambitious project when announced by the (then) Department of Health 5 years ago. Since then, Genomics England and NHS England (now joined by Scotland, Northern, Ireland and Wales), working with a huge number of ground-breaking partnerships, have built the infrastructure and protocols to deliver health-enhancing diagnostics from consented patients with undiagnosed rare genetic disease and common cancers, while at the same time enabling their data (in de-identified form) to provide the basis for research leading to improved therapies and treatments.
>
> Having built the platform and reached the 50,000 halfway point we are now able to operate at a scale to complete the target by the end of 2018.

Currently, the average rare disease patient in the UK consults 5 doctors, receives 3 misdiagnoses and waits 4 years before receiving their final diagnosis.

Genomic testing is changing the lives of patients with a rare disease – often providing diagnoses for the first time after years of uncertainty and distress, known as the ‘diagnostic odyssey’.

Professor Sue Hill OBE, Chief Scientific Officer for England and Senior Responsible Officer for Genomics at NHS England, said:

> This is an important milestone for the project and has only been possible because of the contribution and commitment of the participants in the project and their families.
>
> The milestone also marks how healthcare professionals from across the NHS have come together to transform care for the future, demonstrating how this technology can be utilised as part of routine care to improve patient lives and keep the NHS a world-leader in this important area of medicine. Working together patients and professionals have achieved so much and I would like to say a personal thank you to each and every one for playing their part.
>
> We are on track to complete recruitment to the project this autumn and, from then, the use of these cutting-edge genomic technologies will be embedded in the NHS through the new Genomic Medicine Service offering real benefits to patients and healthcare delivery.