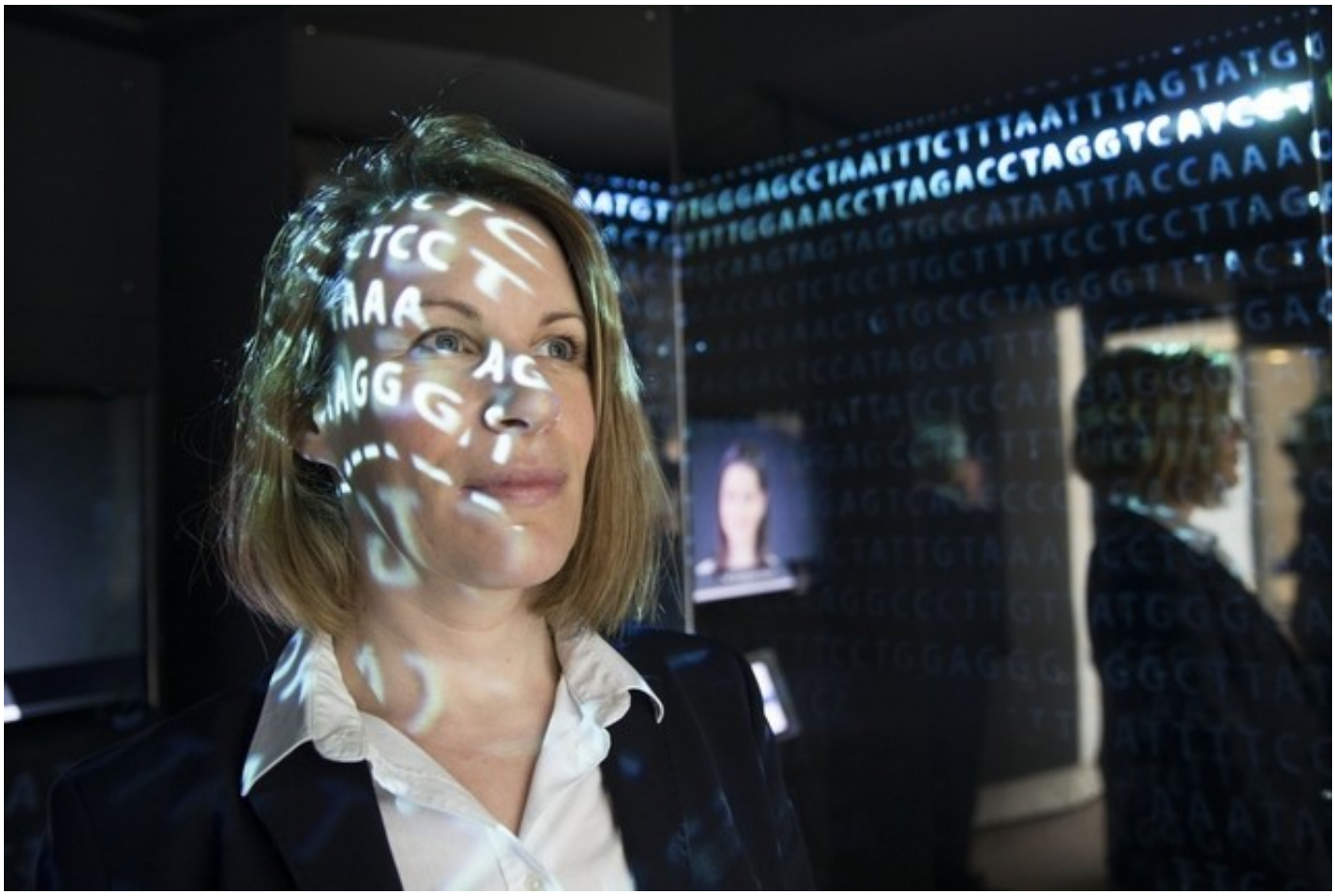

Dr Anna Middleton, who is working on a public engagement project for Genomics England, says the 100,000 Genomes Project research is pioneering and has huge implications for the NHS and patients. Stephen Lock for the National

Gene genie almost out of the bottle

Angela Jameson

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LONDON // Have you ever wondered what your DNA could tell you? Well, soon there will be an app for that.

The journey into the human genome is taking further great strides and the United Kingdom and its public healthcare system, the NHS, is right at the forefront.

A gene is a distinct stretch of DNA that determines something about who you are. Genes vary in size, from just a few thousand pairs of nucleotides (or "base pairs") to more than two million base pairs.

Genomics is a relatively new branch of science which uses DNA sequencing techniques and bioinformatics to sequence, assemble and analyse the function and structure of genomes (the complete set of DNA within an individual).

Looking at a person's genetic make-up can help to assess whether they are at risk from certain diseases and find the best course of treatment for them or suggest lifestyle changes.

UAE among pioneers of a biological frontier

This amazing science has only recently become possible due to an exponential decline in gene sequencing costs, faster processing techniques and large-scale government and pharmaceutical industry investment.

The science is stimulating a potentially huge new life sciences industry as well as major changes in health services.

According to a report by Deloitte for the UK Office of Life Sciences in October, the global genomics market is currently valued at about £8 billion (Dh42.19bn).

Analysts believe the market will grow rapidly, at about 15 per cent a year, as the cost of sequencing gets cheaper and governments and big pharmaceutical companies put huge investment into this field.

Sequencing a human genome now costs just US$1,345 compared with the $95 million it cost in 2001, according to the US National Human Genome Research Institute.

In the UK, the 100,000 Genomes Project was launched in 2012 with £200m of public money and is at the forefront of international research in this area. The project will sequence around 100,000 genomes from 70,000 people with rare diseases and cancer, and their families.

Dr Anna Middleton, the principal social scientist at the Wellcome Trust's Sanger Institute, who is working on a public engagement project for Genomics England, says the 100,000 Genomes Project research is pioneering and has huge implications for the NHS and patients.

"Treatment costs could be reduced but we are also taking away the diagnostic odyssey that some parents endure. After waiting [in the past] years for a diagnosis for their child, a whole genome sequence may offer this in a matter of months," she says.

The NHS is opening 11 Genomic centres across the UK which will give patients better access than ever to testing. But the test is nothing, Dr Middleton says, without sophisticated ways to interpret it. Clinical geneticists and bioinformatics experts are working hard on this and genetic counsellors, of which there are just 300 in the NHS, have to explain what the results mean in practical terms.

The 100,000 Genomes Project should usher in a new era of personalised medicine but it should also generate research and business opportunities. For a start, Deloitte predicts that it will generate demand for formalised data interpretation companies.

Tom Slater, a fund manager at the Scottish firm Baillie Gifford has been investing in the genomics sector for five years, largely through the US company Illumina, which is a market leader in selling the machines that conduct genomic sequencing. "The possibilities that cheap genetic sequencing throws up are incredibly important for improving health care outcomes and taking cost out of health systems," he says. But he points out that there are still big challenges to overcome, particularly when it comes to the issue of collecting and sharing data.

Because the volumes of data are so huge, many analysts expect the big tech innovators – such as Google and Amazon – to become involved in this area, because they have the computing power and expertise to process

such vast amounts of information. It is only by processing data on a huge scale that health professionals can begin to draw useful diagnostic patterns and predictors that can improve people's chances of beating diseases.

Last month, one of the UK's biggest pharmaceutical companies AstraZeneca said it would invest "hundreds of millions of dollars" over the course of 10 years, as it launched a huge effort to compile genome sequences and health records from two million people.

Astra says it will use the data to help drug development in all of its major disease areas, from diabetes to inflammation to cancer. It is partnering research institutions including the Wellcome Trust Sanger Institute, and Human Longevity, a biotechnology company founded in San Diego, California, by the genomics pioneer Craig Venter.

AstraZeneca also expects to draw on data from 500,000 participants in its own clinical trials, and medical samples that it has accrued over the past 15 years. GSK, the UK's biggest pharma company, is investing £65m over the next five years in a new non-profit research centre in Seattle called the Altius Institute for Biomedical Sciences. GSK wants to better understand biology so it can discover more medicines, like every other drug maker. It also wants to quit wasting money on drug candidates that look promising in the lab but flop years later when given to thousands of real people.

GSK is betting that one way around the problem will come from "the living genome" or what some call the "dark matter" of the genome. These mysterious stretches in the genetic instructions do not contain genes that provide code for making proteins, but they do appear to provide important controls over what genes do in different cells, in different states of health and disease, and in response to different environments.

But collecting data on such an enormous scale raises significant ethical questions, which Dr Middleton is considering in two research projects. A project called www.YourDNAYourSay.org looks at attitudes towards big data and genetics, while if you wondered how you might start a conversation about genomics – and experts are certain an increasing number people will soon want to do so – then a series of amusing short films on www.GeneTube.org should help you to get a grip on the basics.

One of the reasons it is important that the wider public understand how their personal data is being collected is that lots of commercial companies are keen to do it, too. For instance, Apple has developed a software platform that helps hospitals or scientists run medical studies on iPhones by collecting data form the devices' sensors or through surveys.

Google also has a little-known life sciences spin-off called Verily. The company is now independent of the search giant, but its website has moved away from its original life sciences brief and says it is now focused on using technology to better understand health, as well as prevent, detect and manage disease.

Back in the UK, there is talk that a gene-sequencing specialist that was spun out of Oxford University 11 years ago could list on the London stock exchange this year, with a market value of up to £1bn. Oxford Nanopore Technologies has developed a proprietary technology for genetic sequencing. Its two instruments – GridION, for large scale projects, and MinION for smaller ones. The MinION is the size of a memory chip. Since its launch in 2005, Oxford Nanopore has attracted £251m of funding and its big

investors include the listed fund IP Group and the star fund manager Neil Woodford's Woodford Investment Management.

Meanwhile, the Cambridge university professor who invented the world's most successful technology for reading DNA has secured $29m in financing for his latest venture. Shankar Balasubramanian is seeking to exploit epigenetics – nature's instructions for switching genes on and off. He founded Solexa in 1998, which was acquired for $650m in 2007 by Illumina, which used its technology to become the market leader for reading genes.

The potential of gene editing has pushed more than $1bn of venture capital investment into gene-editing companies in the past two years, according to Boston Consulting Group.

Crispr, as the editing technology is known, is already being used by British scientists to redesign livestock, including editing cattle DNA to stop them growing horns.

AstraZeneca has invested millions in Crispr and is using it to edit the genomes of mice and of human cells to pursue therapies for heart disease, cancer and other illnesses. It believes Crispr could slash billions off the pharma industry's research and development costs. It is hoped this, in turn, would be reflected in the prices of new medicines.

Novartis, which has signed two deals with the gene-editing start-ups Intellia Therapeutics and Caribou Biosciences, plans to use Crispr for engineering immune cells and blood stem cells, and as a research tool for drug discovery.

Editas Medicine, which is looking at rare eye disease, raised $95m through a float in the US in February, while Intellia raised $108m in its own IPO this month. It develops treatments for cancer and liver disease.

Darren Griffin, a professor of genetics at the University of Kent, says the new treatments offer real hope: "Crispr technology is offering a range of exciting applications including treatment options for genetic diseases."

Silence Therapeutics is a UK biotechnology company that develops gene therapeutic technology based on RNA interference. RNA is the molecule responsible for controlling gene expression in nature. Gene expression determines what a cell is able to do.

Institutional investors including Henderson and Aviva have invested in Silence Therapeutics, but even its most advanced treatment – for pancreatic cancer – is still only in a clinical trial.

While it could still be years before investors see the sort of returns they hope for, few in the field doubt the coming genetics revolution in health care and its wider applications will affect every aspect of life, potentially changing humankind forever.

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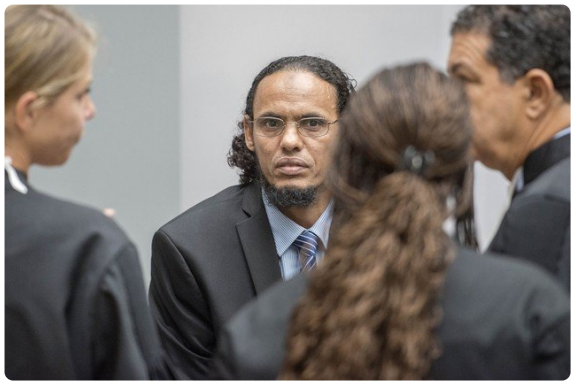

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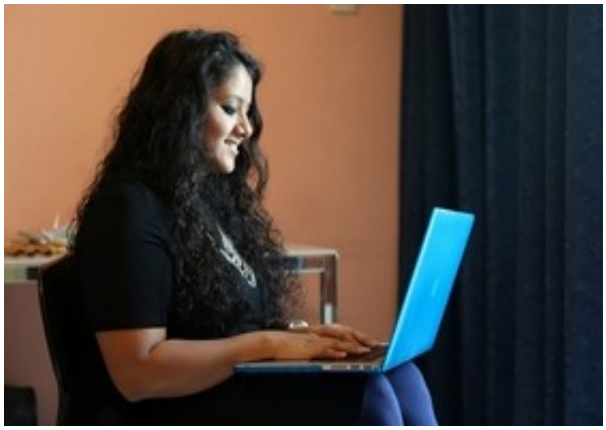

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