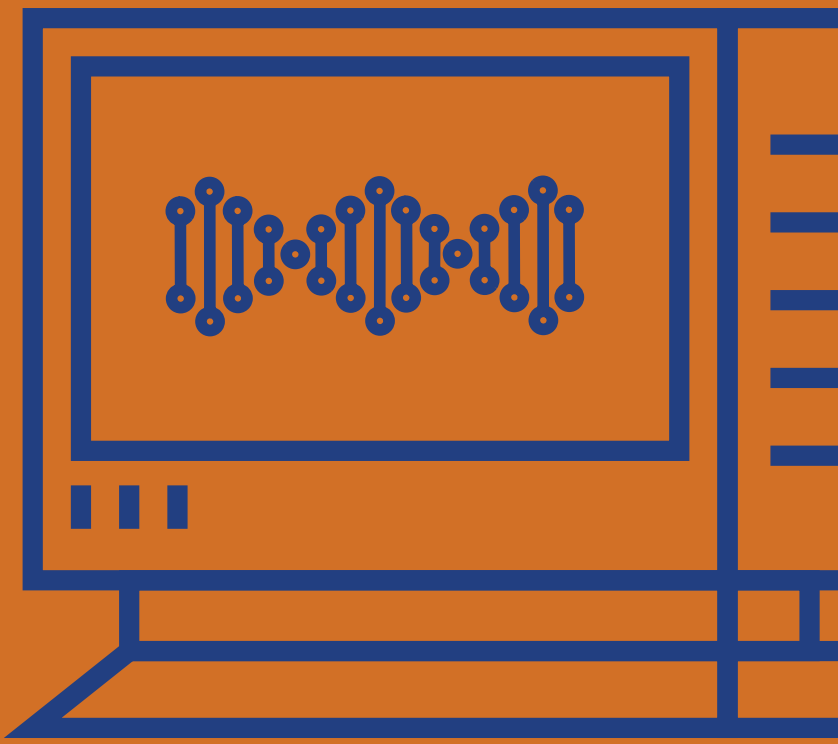


GENOMICS EXPLAINED

A summarised guide to genomics

UK BioIndustry Association

2019

Foreword

The 21st century is proving to be one of the most exciting and prolific periods of innovation in biosciences and healthcare, and UK bioscience companies are at the forefront of this innovation.

These companies are a key part of the UK Bioindustry Association (BIA)'s membership, and we provide a home for them through our Advisory Committees and working groups on antimicrobial resistance, cell and gene therapy, engineering biology and genomics.

We are delighted to publish this short summary of **A guide to genomics and UK excellence in the field** as an accompaniment to our Explainer series, within which we describe what these four strategic technologies are all about, and showcase the important contributions being made by some of the UK bioscience firms who make up our dynamic and innovative membership. You can access the full versions of these Explainer documents on our website, or get in touch with us if you would like some hard copies.

We hope you enjoy reading them.

Steve Bates OBE CEO,
UK Bioindustry Association

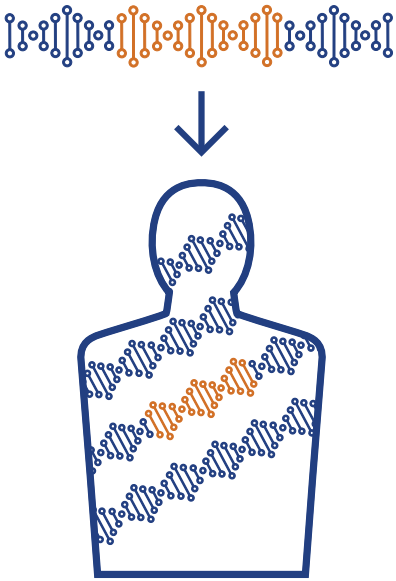
What is genomics?

At the start of the 21st century, the Human Genome Project unravelled the human genetic code, which was a ground-breaking piece of work. But learning how to interpret and, in some cases modify, that genetic code is even more significant. That's genomics – reading, understanding and editing the genetic instructions, or DNA, that make up each one of us. DNA is a chain of four types of molecule, called nucleotides, arranged in the iconic double-helix structure. Each nucleotide is represented by a letter – A, G, C or T. A gene is a specific sequence of these four letters, whose arrangement contains instructions to make a particular protein. These proteins determine what we look like, how we develop and function.

Human DNA contains about 20,000 genes. When cells divide and multiply, errors can occur as the long, highly-repetitive DNA code is copied. These coding errors, known as 'mutations', can lead to disease. The mutations may be inherited, or caused by environmental factors, such as exposure to toxins or radiation. Some diseases, like cystic fibrosis, are associated with just one faulty gene, occurring when children inherit two copies of a faulty gene called cystic fibrosis transmembrane conductance regulator (CFTR). Other diseases, like many cancers, are associated with mutation across lots of different genes. Genomics is a new frontier of health research, working to unravel these complex problems.

Wellcome Genome Campus

The Wellcome Genome Campus is home to research institutes, spin-outs and start-ups, academic-industry partnerships and Genomics England; all dedicated to driving and leading genomics research and innovation. The **Wellcome Sanger Institute**, a major contributor to the Human Genome Project, is built to think big. The Wellcome Sanger Institute sits next to one of the biggest life sciences data repositories, EMBL's European Bioinformatics Institute. This innovation and data-rich infrastructure is expanding, for instance, the BioData Innovation Centre opened in 2016 at the Wellcome Genome Campus to house emerging genomics and biodata companies.



Congenica

Among those emerging companies is Congenica, whose genome-analysis software filters raw genomic data and clinical information to help clinicians diagnose and treat patients with inherited diseases. Their Sapientia platform is designed to provide genome sequencing, annotation, interpretation and customisable clinical reports. Sapientia interprets this information to rapidly deduce the most accurate diagnosis. Sapientia is being used by the NHS to make important diagnoses and improve the lives of patients. In China, it is supporting whole-genome sequencing as part of the country's 100k Wellness Pioneer project, seeking to improve health through understanding the genomes gathered from 100,000 Chinese citizens.

Genomics plc

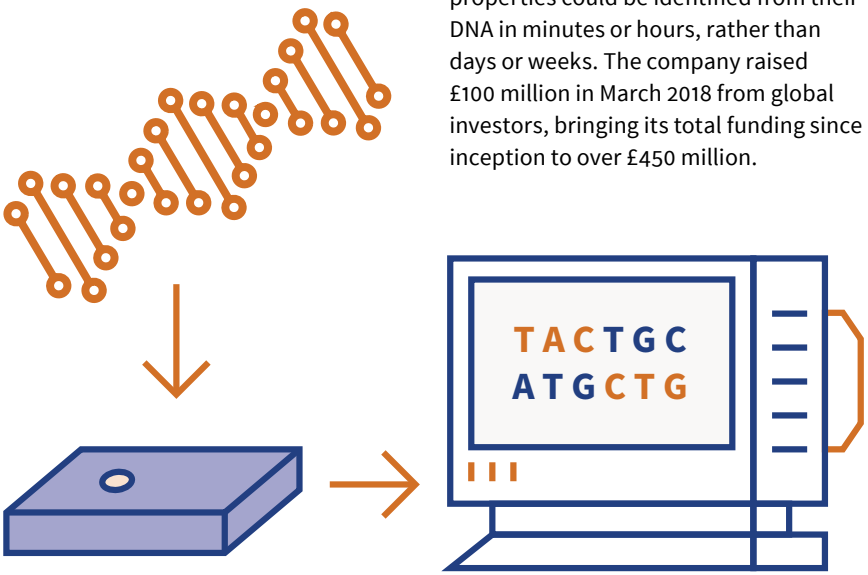
Oxford-based Genomics plc's mission is to turbo-charge drug discovery by better understanding human biology and disease. The company has built one of the largest repositories of genomic and phenotypic information, capturing the effects of millions of genetic variants on health and disease. Genomics plc applies machine learning to these vast databases, seeking novel genetic and molecular pathways that may form the basis of new drugs. In October 2017, US biotech giant Biogen signed up Genomics plc to help find new drug targets for multiple sclerosis.

Global Gene Corp

Global Gene Corp. is building longitudinal genomic data sets from under-explored populations in India and South Asia. The genomics data company is working with over 50 partners worldwide to help democratise precision medicine, by ensuring that data is collected from less-studied patient populations. At the moment, 60% of the world's population accounts for only a tiny portion – less than 5% – of available genomics data. Yet certain diseases, such as cancer, may manifest differently in Indian and South-east Asian patients versus those in the US or Europe. Understanding those differences may ultimately lead to better treatments for patients everywhere. Global Gene Corp. has R&D facilities at the Wellcome Genome Campus in Cambridge, UK and offices in Boston and Singapore.

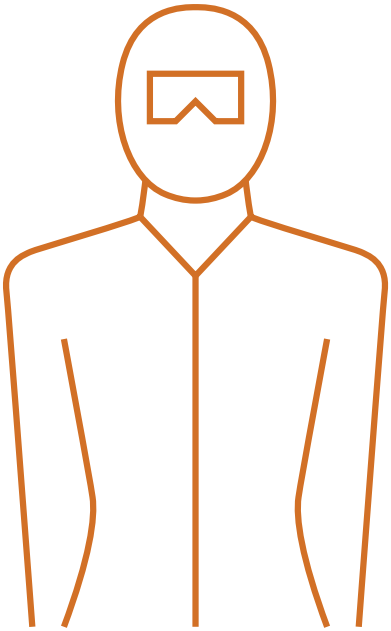
Oxford Nanopore

Oxford Nanopore's range of real-time, low-cost DNA sequencing devices, from benchtop to pocket-sized, are opening up biological analyses to scientists across multiple disciplines, in multiple locations. Its tiniest sequencer, which weighs less than 100g and plugs into a laptop, has been used to answer biological questions on farms and in food production factories, and even on the International Space Station. The technology involves passing currents through biological nanopores and measuring changes as molecules, like DNA or RNA, pass through or close to the nanopore. As a sample is analysed, data is streamed in real-time, meaning researchers can stop as soon as they have enough data to answer their question. Such rapid results mean that, for example in infectious diseases, pathogens and their drug resistance properties could be identified from their DNA in minutes or hours, rather than days or weeks. The company raised £100 million in March 2018 from global investors, bringing its total funding since inception to over £450 million.



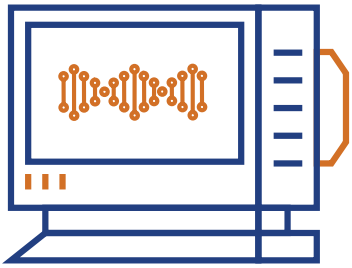
Cambridge Cancer Genomics

Cambridge Cancer Genomics is using artificial intelligence and smart genomics to develop new, smarter blood tests that can rapidly signal whether, and how well, a patient's cancer treatment is working. Liquid biopsies test the blood for cancer cells or cancer cell DNA that may be circulating in the blood. Besides being relatively quick and simple, liquid biopsies could help pick up cancer at an earlier stage than other kinds of tests. Liquid biopsies may one day be able to better predict the most appropriate therapy for patients before treatment has even started.



Cambridge Epigenetix

Cambridge Epigenetix is building diagnostics and therapies based on their understanding of the multiple epigenetic influences on disease. The epigenome is the set of chemicals and processes that make up our genetic "control panel", regulating which genes are turned on or off, when, and for how long. Multiple external factors can affect the structure of our epigenome, and with that, our risk of disease. Cambridge Epigenetix's range of technologies enable the identification of sensitive, disease-specific epigenetic markers from samples of blood and tissue.



Such epigenetic markers may enable earlier detection of certain complex diseases like cancer. The company is developing diagnostics for various important diseases and offers a biomarker discovery service. The company is privately owned and was spun out of the University of Cambridge in 2012; its backers include Google Ventures and Syncona.

To access the full version of this report, take a look at our Strategic Technologies page: www.bioindustry.org/policy/strategic-technologies.html

This report would not have been possible without the input and expertise from BIA member companies, who we thank for their contributions. The production of this report was also supported by life sciences specialist writer Melanie Senior, who we also acknowledge and thank for her contribution to the content of this report.

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