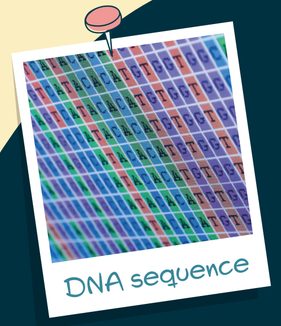
Unravelling the genome

Understanding the human genome can be seen as the key to understanding the mystery of human life itself. Since the first complete sequence of the human genome was revealed in 2003, things have moved fast. Advances in bioinformatics and big data gathering and handling mean timescales are changing all the time. It took years to produce the first sequence of human DNA. Now it is vastly quicker to sequence the entire human genome. Scientists are taking advantage of these developments to delve ever deeper into what makes us human – and what keeps us healthy.



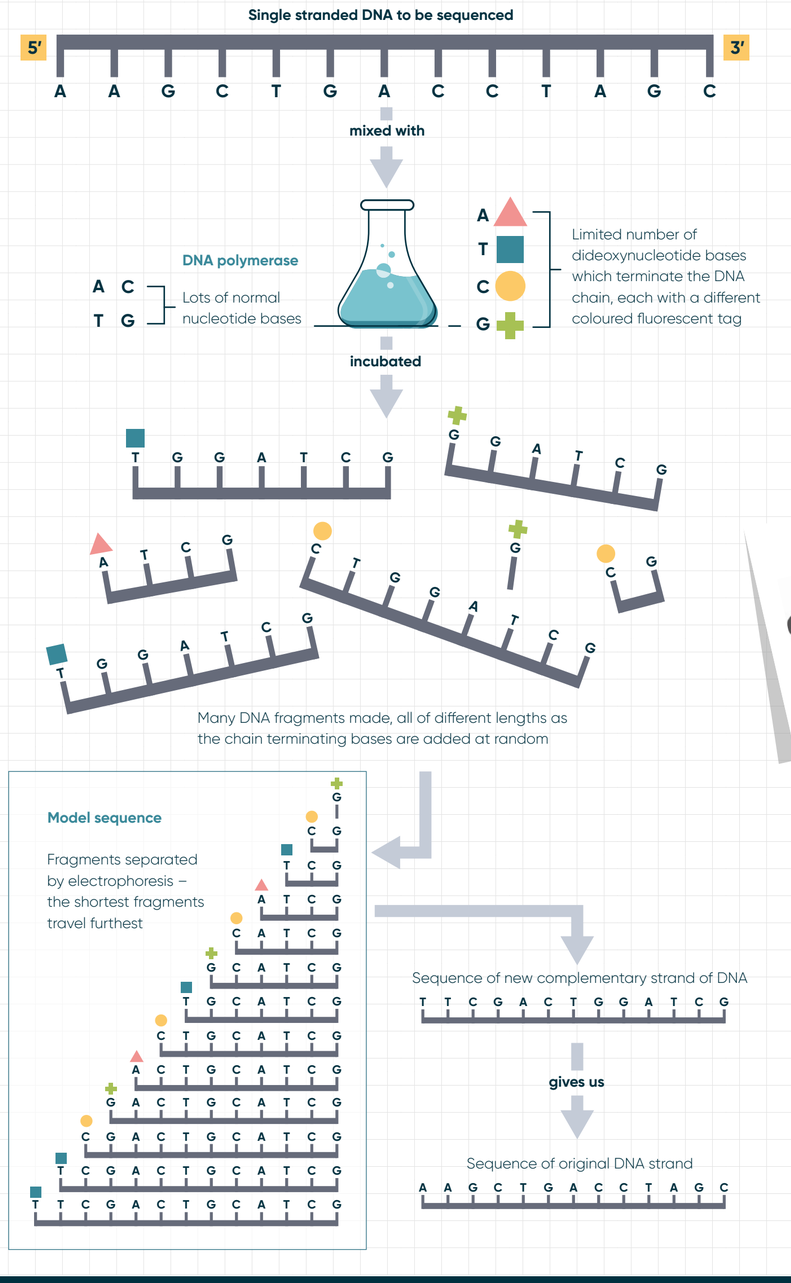
The Human Genome Project

The Human Genome Project (HGP) set out to identify all of the genes in the human chromosomes (around 30,000 of them) and to sequence the billions of base pairs which make up human DNA. The HGP was an example of successful international cooperation, with scientists in 18 countries all working on sequencing the DNA and sharing their results. As a result of advances in the technology used to sequence DNA, the task of producing the complete human genome was completed two years ahead of schedule and under budget!

New sequencing methods

The basic principles of DNA sequencing remain the same, but the technology has come a long way since Frederick Sanger and his team first sequenced the genome of the virus phiX174 and the science of genomics was born. Automation, orders of magnitude more computing power and changes to the process have resulted in new generations of sequencing machines. Massively parallel sequencing involves sequencing thousands of clusters of DNA at the same time. As a result, we can sequence the 3 billion bases of the human genome in a matter of days and at a fraction of the original cost. And it's getting faster and more affordable all the time.

The method of DNA sequencing used in the Human Genome Project



1000 Genomes Project

Between 2008 and 2015 scientists ran the 1000 Genomes Project. This used new sequencing technologies to analyse the genomes of at least a thousand anonymous people from different ethnic groups. It was another massive international project, with scientists in countries including the UK, the US, Nigeria, China, Japan, Italy and Peru all sharing data. The aim was to build up a huge catalogue of human variation, to help us understand more about both human health and disease. It brings the prospect of truly personalised medicines closer.

100K Genomes Project

This UK project, using the latest DNA sequencing technologies, aimed to sequence 100,000 genomes from NHS patients. The 100,000 Genomes Project focused mainly on patients with rare diseases and their families, and patients with different types of cancers. This massive undertaking aimed to throw light on the genetic variations which lead to disease and to change the way we identify and treat cancers. The 100,000 genomes were sequenced by the end of 2018, but research and analysis is still ongoing, and provides another step towards a future of personalised medicines.

Sequencing in the surgery

Reading the genome makes many things possible, from pinpointing people at high risk of developing certain diseases to picking up cancers as early as possible. Personalised medicine can become a reality, enabling doctors to choose the most effective drugs for each individual patient. Reading the genome of a pathogen means doctors can offer the right antibiotic – or none at all. In future, perhaps every hospital and GP practice will have easy access to the technology needed to sequence key areas of both patient and pathogen DNA, making genomics a part of routine healthcare.

Use QR code to find out more about unravelling the genome on the ABPI Schools website

www.abpischools.org.uk

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