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New maps for navigating the genome unveiled by scientists

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Scientists have built the clearest picture yet of how our genetic material is regulated in order to make the human body work.

They have mapped how a network of switches, built into our DNA, controls where and when our genes are turned on and off.

University of Edinburgh scientists played a leading role in the international project – called FANTOM5 – which has been examining how our genome holds the code for creating the fantastic diversity of cell types that make up a human.

The three year project, steered by the RIKEN Center for Life Science Technologies in Japan, has involved more than 250 scientists in more than 20 countries and regions.

The study is a step change in our understanding of the human genome, which contains the genetic instructions needed to build and maintain all the many different cell types in the body. All of our cells contain the same instructions, but genes are turned on and off at different times in different cells.

This process is controlled by switches – called promoters and enhancers – found within the genome. It is the flicking of these switches that makes a muscle cell different to a liver or skin cell.

The team studied the largest ever set of cell types and tissues from

human and mouse in order to identify the location of these switches within the genome. They also mapped where and when the switches are active in different cell types and how they interact with each other.

Today the consortium publishes a series of papers describing its findings, including a pair of landmark papers in the journal *Nature*.

In a separate study, researchers at the University of Edinburgh's Roslin Institute have used information from the atlas to investigate the regulation of an important set of genes that are required to build muscle and bone. Another study has used the atlas to investigate the regulation of genes in cells of the immune system.

The FANTOM5 project included major contributions from The Roslin Institute, which is funded by the Biotechnology and Biological Sciences Research Council, and the Medical Research Council Institute of Genetics and Molecular Medicine at the University of Edinburgh.

Professor David Hume, Director of The Roslin Institute and a lead researcher on the project, said: "The FANTOM5 project is a tremendous achievement. To use the analogy of an aeroplane, we have made a leap in understanding the function of all of the parts. And we have gone well beyond that, to understanding how they are connected and control the structures that enable flight.

"The FANTOM5 project has identified new elements in the genome that are the targets of functional genetic variations in human populations, and also have obvious applications to other species."

Dr Martin Taylor, from the MRC Institute of Genetics and Molecular Medicine at the University of Edinburgh, said: "The research gives us an insight as to why humans are different from other animals, even though we share many genes in common. Comparing the mouse and human

atlases reveals extensive rewiring of gene switches that has occurred over time, helping us to understand more about how we have evolved."

The researchers are now establishing the new technology developed in Japan at the University of Edinburgh's high-throughput genomics facility, Edinburgh Genomics, to enable rapid access for researchers based in the UK.

More information: dx.doi.org/10.1038/nature13182
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