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DNA study could shed light on how genetic faults trigger disease

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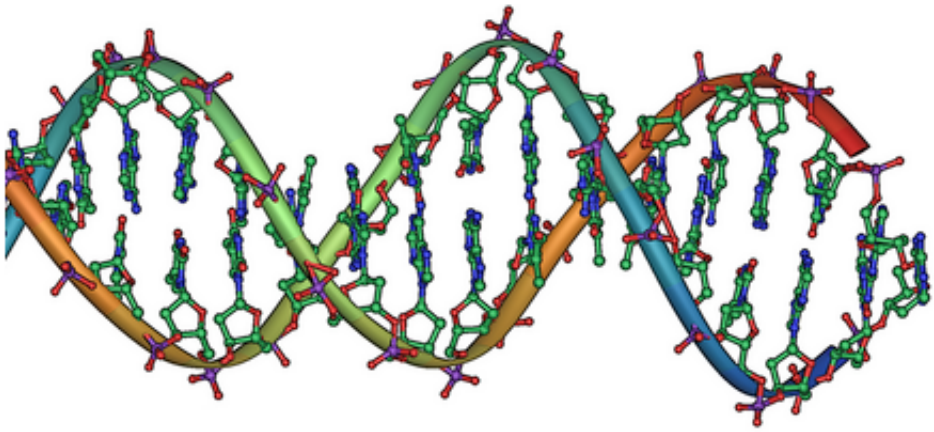

DNA double helix. Credit: public domain

A new technique that identifies how genes are controlled could help scientists spot errors in the genetic code which trigger disease, a study suggests.

The method focusses on those parts of DNA - known as enhancer regions - which regulate the activity of genes and direct the production of proteins that have key functions within the body.

Errors in protein production can result in a wide range of diseases in people, researchers say.

The new method could help researchers pinpoint the source of disease-causing mutations in enhancers. Until now, these genetic errors have been difficult to interpret as the link between enhancers and the genes they control was not clear.

Researchers at the University of Edinburgh were part of an international collaboration that identified all the enhancers - and the genes they activate - on a single human chromosome.

The team then tested the technique in zebrafish and found that genes are controlled by enhancers in a similar way, suggesting that this type of regulation takes place in all animals.

Individual genes may be under the control of many enhancers, which allow gene activation to be carefully regulated, the team says. This allows precise control of gene activity, which is important during development and in maintaining normal brain function.

The study, published in the journal *Nature Communications*, was funded by the Seventh Framework Programme of the European Union. The study was carried in close collaboration with researchers based in other parts of the UK, France, Germany, Australia, and Norway.

Professor David FitzPatrick, of the University of Edinburgh's MRC Human Genetics Unit, who took part in the study, said: "This work is an important step in identifying which enhancers control which genes, and this will help us in interpreting the genetic changes we see in the part of the genome that does not code for protein."

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Provided by University of Edinburgh

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