

Genes that only humans have

Michael Le Page

AT LEAST three human genes evolved “from scratch” via mutations in non-coding stretches of DNA, a process thought to be virtually impossible until recently. The genes evolved since human and chimp lineages split and so are unique to us.

It is tempting to speculate that uniquely human genes drive uniquely human traits, says Aoife McLysaght of the Smurfit Institute of Genetics at Trinity College Dublin, who discovered the three genes with her colleague David Knowles. But that would be premature: “There is no clue about the function at all yet.”

What is extraordinary about the genes is their evolutionary past. Most new genes arise when existing ones are duplicated and the copies slowly acquire different functions. The three new genes, called *CLLU1*, *C22orf45* and *DNAH10OS*, suddenly sprang into existence as a result of mutations in DNA sequences that did not previously code for proteins.

While comparing human, chimp and macaque DNA, McLysaght’s team stumbled upon human genes that stood out

because they seemed to have no equivalents in the other species. The short proteins the three genes code for have been found in blood samples from healthy people, and the genes are also present in all human genomes sequenced so far, suggesting they have vital functions in humans.

In the other primates, the equivalent DNA sequences contained differences that would halt protein production early on, so the sequences are non-coding in these species. Crucially, chimps, gorillas, gibbons and macaques share some of these differences, meaning that, in our shared ancestor, these sequences were non-coding as well.

The researchers conclude that three of these non-coding

“The three new genes seem to have arisen as a result of mutations in non-coding sequences of DNA”

sequences must have mutated in humans and become capable of coding for the short proteins at some point since we diverged from chimps six million years ago (*Genome Research*, DOI: 10.1101/gr.095026.109).



Six million years later

Such “de novo” gene evolution was once thought impossible because random mutations are highly unlikely to produce a DNA sequence that encodes a protein of any length, let alone a protein that will be transcribed by cells and do anything useful. But in 2006, several de novo genes were discovered in fruit flies. Since then, it’s become clear that genes do continually evolve in this way.

Part of the explanation might be that biological systems are very noisy: even though most of our DNA is junk, most of it still gets transcribed into RNA at times, and some of that RNA probably

reaches cells’ protein-making machinery. This means that when mutations do throw up sequences capable of encoding proteins, some may get “tested” and useful ones selected for. As more primate genome data becomes available, McLysaght estimates a further 15 human genes will turn out to have evolved de novo.

She now plans to find out what the three genes she found do, but it won’t be easy. The conventional method for discovering what genes do is to disable them in mice to see what the effect is – the trouble is that no animals besides humans have these genes. ■

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