

M1 Cours de traduction scientifique et technique : M. Benayoun et Mme Minacori

<http://www.economist.com/node/21562185/print>

The nature of man. Large-scale genetic studies are throwing light on what makes humans human

Sep 8th 2012 | from the print edition

HUMANS are peculiar as a species, so what makes them so must be hidden in their genome. To an almost disconcerting extent, though, the human genome looks similar to the genomes of other primates, especially when it comes to the particular proteins it allows cells to make. The powerful new ways of looking at the genome being pioneered by the ENCODE consortium (see [article](#)), though, provide ways to seek out the subtle species-specific signals. Lucas Ward and Manolis Kellis of the Massachusetts Institute of Technology report on the results of such sleuthing in a paper just published in *Science*.

The two researchers used data from ENCODE to identify the bits of the genome that actually do things and data from the 1,000 Genomes Project, which has studied human-genome variation across hundreds of people, to discover how much these functional elements vary from person to person. In particular, they looked for telltales that an element is being maintained by natural selection. If something is evolutionarily important then random variations in its DNA sequence will be slowly eliminated from the population, keeping it on the functional straight and narrow in a process known as purifying selection.

Dr Ward and Dr Kellis found that, in addition to the 5% of human DNA that is conserved between mammals, an additional 4% of human DNA appears to be uniquely human in the sense that it is prone to purifying selection in humans but not in other mammals. Much of this proprietary DNA is involved in regulating gene activity—for example, controlling how much of a protein is produced, rather than changing the nature of the protein itself. This finding is in line with modern thinking that a lot of evolutionary change is connected with regulatory elements rather than actual protein structure. The researchers also found that long non-coding segments that are not conserved in other mammals are in fact highly constrained in humans, suggesting they have human-specific functions.

Some areas identified as particularly human are the regulation of the cone cells of the retina (which are involved in colour vision) and the regulation of nerve-cell growth. These processes evolved rapidly in man's primate ancestors but are now under strong purifying selection to maintain their beneficial functions. The implications of that, given humanity's main distinguishing feature—its huge brain—are obvious. Dr Ward and Dr Kellis have thus created a powerful tool for investigating in detail just what it is that makes a human being human.

from the print edition | Science and technology