

The genetic footprint of natural selection

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A further step has been taken towards our understanding of natural selection. CNRS scientists working at the Institut de Biologie of the Ecole Normale Supérieure (CNRS, February) have shown that humans, and some of their primate cousins, have a common genetic footprint, i.e. a set of genes which natural selection has often tended to act upon during the past 200,000 years. This study has also been able to isolate a group of genes that distinguish us from our cousins the great apes. Its findings are published in *PloS Genetics* (26 February 2010 issue).

During evolution, living species have adapted to environmental constraints according to the mechanism of natural selection; when a mutation that aids the survival (and reproduction) of an individual appears in the genome, it then spreads throughout the rest of the species until, after several hundreds or even thousands of generations, it is carried by all individuals. But does this selection, which occurs on a specific gene in the genome of a species, also occur on the same gene in neighboring species? On which set of genes has natural selection acted specifically in each species?

Researchers in the Dynamique et Organisation des Génomes team at the Institut de Biologie of the Ecole Normale Supérieure have studied the genome of humans and three other primate species (chimpanzee, orangutan and macaque) using bioinformatics tools. Their work consisted in comparing the entire genomes of each species in order to identify the genes having undergone selection during the past 200,000 years. The result was that a few hundred genes have recently undergone selection in each of these species. These include around 100 genes



detected in man that are shared by two or three other species, which is twice as many as might be anticipated as a random phenomenon. Thus a not inconsiderable proportion of the genes involved in human adaptation are also present in the chimpanzee, orangutan or macaque, and sometimes in several species at the same time. <u>Natural selection</u> acts not only by distancing different species from each other when new traits appear. But by acting on the same gene, it can also give rise to the same trait in species that have already diverged, but still have a relatively similar genome.

This study thus provides a clearer understanding of the group of genes that are specifically implicated in human evolution (during the past 200,000 years), as it allows the identification of those genes which did not undergo selection in another primate line. An example that has been confirmed by this study is the well-known case of the lactase gene that can metabolize lactose during adulthood (a clear advantage with the development of agriculture and animal husbandry). The researchers have also identified a group of genes involved in some neurological functions and in the development of muscles and skeleton.

The degree of variability as a selection indicator

Until now, the identification of selected genes required the study of the genomes of several dozens of individuals based on statistical methods and had only been performed in humans. The CNRS researchers have developed a method that only requires the genome of a single individual. This is based on searching for genomic regions with very little allele polymorphism. Two copies of each gene are present in the genome and are referred to as alleles (one on each chromosome), and they are not perfectly identical; a certain degree of polymorphism is present. When an advantageous mutation occurs and then spreads throughout the population, the genome of each individual becomes identical in the region surrounding the gene concerned. In this case, polymorphism is



very weak: an advantageous mutation has been selected to the detriment of local variability in the <u>genome</u>.

Using a larger number of primate genomes, the study now needs to determine the extent of this phenomenon in terms of <u>genes</u> and biological functions. By including other vertebrate species in the study, it will also be possible to determine whether we share adaptive events with rodents, birds or fish, as some isolated observations appear to suggest.

More information: Enard, D., Depaulis, F., Roest Crollius, H. (2010) Human and non-human primate genomes share hotspots of positive selection. *Plos Genetics* 6(2): e1000840. <u>doi:10.1371/journal.pgen.1000840</u>

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