

120<sup>ème</sup>  
anniversaire

Institut Pasteur



Paris, 4 February 2008

Press release

## **Natural selection in the human genome: new leads for understanding our predispositions to diseases**

**Researchers from the Institut Pasteur and the CNRS published a study of human population genetics in *Nature Genetics*, conducted across the full scope of the genome. It allowed the identification of a group of over 580 genes that have very likely contributed to the morphological diversity of populations, and to their differences with respect to disease susceptibility. The researchers' work opens important investigative leads in the latter domain for the study of genes that predispose for different pathologies.**

Are the differences in hair colour, height, or susceptibility to certain diseases observed between different human populations today the result of chance (genetic drift), or natural selection—in other words, the process of adapting to the environment? And if natural selection played a role in the phenotypic differences between populations, which genes were specifically involved?

Researchers from the Human Evolutionary Genetics Unit (CNRS URA 3012), directed by Lluís Quintana-Murci at the Institut Pasteur, sought to answer these questions by conducting a wide-ranging analysis across the genome. It was performed using data generated by the international project HapMap, the objective of which is to develop a public resource to aid researchers in discovering the genes linked with human diseases and with responses to medication.

The analysis involved 210 individuals representative of different human populations. The level of differences between populations was measurable using more than 2.8 million polymorphic markers of the human genome.

The study made it possible to prove that natural selection played a significant part in the diversity of modern human populations. Further, the study led to the identification of 582 genes, which, having been subjected to strong pressures of positive selection, vary between human populations. These genes would have become differentiated between 60,000 and 10,000 years ago.

Some of them are involved in physical differences (skin pigmentation, hair thickness), others play a role in host-pathogen interactions—such as gene CR1, implicated in the severity of malaria attacks, a mutation of which is found in 85% of Africans, but lacking in Europeans and Asians. Finally, other genes are linked with diseases for which the prevalence rate may vary between populations, such as diabetes, obesity, or hypertension.

*"These genes have contributed to adaptation to the environment; their mutations have provided selective advantages: thus they probably play an important role on the scale of the individual", stressed Quintana-Murci. He clarified: "However, these genes explaining phenotypic differentiation between populations only represent a minute portion of our genome, which again abolishes the concept of 'race' from a genetic point of view."*

*"Our work therefore opens the way to medically significant genetic research, in particular by having identified candidate genes for diseases, the prevalence of which varies between human populations, and candidate genes for the predisposition to different pathologies", the researcher concluded.*

---

**Source:**

*"Natural selection has driven population differentiation in modern humans", **Nature Genetics**, AOP 3 February 2008.*

Luis B. Barreiro, Guillaume Laval, H el ene Quach<sup>1</sup>, Etienne Patin, and Lluis Quintana-Murci

Human Evolutionary Genetics Unit (CNRS URA 3012)

---

**Contact persons:**

- Press Office, Institut Pasteur: Corinne Jamma or Nadine Peyrolo – +33 (0)1 40 61 33 41 – [cjamma@pasteur.fr](mailto:cjamma@pasteur.fr)
  - Press Office, CNRS: Muriel Ilous - +33 (0)1 44 96 43 09 [muriel.ilous@cnrs-dir.fr](mailto:muriel.ilous@cnrs-dir.fr)
-