



PRESS RELEASE

A large-scale genetic study confirms that Basque people are a homogeneous group distinguishable from other European populations

- *The project, promoted by a consortium made up of CIC bioGUNE, the University of the Basque Country and the Basque Biobank, has been published in the prestigious journal Human Genetics.*
- *A total of 60,000 genetic markers from 240 European citizens have been used, of which 83 are Basque-Spanish and 24 Basque-French.*
- *This is the first large-scale population genetics study performed on the Basque-Spanish and Basque-French population.*

(Bilbao, May 10, 2010).- A new population genetics study coordinated by the Centre for Cooperative Research in Biosciences ([CIC bioGUNE](#)) confirms that native Basque population, both in Spanish regions and the French Basque Country, is a genetically homogeneous group clearly distinguishable from other European populations, as published by the prestigious journal *Human Genetics*.

The study was conducted by a consortium made up of [CIC bioGUNE](#), the [Department of Genetics, Physical Anthropology and Animal Physiology of University of the Basque Country \(UPV/EHU\)](#) and the Basque Biobank for Research (O+Ehun) of the [B+I+O eusko fundazioa](#).

This project, launched in 2005 by collecting samples, is the first large-scale population study which includes individual genetic data of Basque citizens living in French and

Spanish territories. A total of 60,000 genetic markers from 240 European citizens have been analyzed, of which 83 are Basque-Spanish (22 from each region of the Autonomous Basque Community –Alava, Biscay and Gipuzkoa–, and 17 from the Foral Community of Navarre), and 24 Basque-French. The remaining 133 people are from different European populations (France, Northern Italy, Tuscany, Orkney Islands – Scotland–, Sardinia, Russia and Republic of Adygea –Russia–).

Genetic data for European populations have been obtained from the HGDP-CEPH Human Genome Diversity Cell Line Panel database, belonging to the [Fondation Jean Dausset](#). Otherwise, the samples and genetic data from the Basque-Spanish citizens have been gathered specifically for this study and will be available for future studies.

The samples from the Spanish-Basque citizens categorised in this study form part of the Basque Biobank (O + Ehun) collection.

➤ **Selection of samples**

The criteria for selecting the population base for this study followed very strict guidelines. Each individual giving his/her consent to participate had to corroborate (by civil or ecclesiastical record) that his/her three previous generations (parents, grandparents and great grandparents) were all born in the same province.

Furthermore, people chosen to participate in the study had to prove that they did not suffer from any disease – that is to say, they were healthy. Thus, relevant basic biochemical analysis of blood (cholesterol level, transaminases, etc.) was carried out for all the samples collected. When possible, samples from the parents of those selected were also gathered. Out of the 200 samples that were initially collected, the genetic study group was reduced to 83, which has been the definitive basis on which this research has been carried out.

All individuals who met the established criteria donated 15 ml of blood. For the purposes of the research, the deoxyribonucleic acid (DNA) was extracted from the lymphocytes. All the samples are now available for researchers who require them for other specific scientific studies, provided that the objectives of their use pass the review and control process of the scientific and ethical committees of the [B+I+O eusko fundazioa](#).

"This project required a lot of coordination, both in terms of the collection of samples and the proper interaction of groups from very different backgrounds, as apart from the participation of collaborating groups (BIOEF, UPV-EHU and CIC bioGUNE), certain services were subcontracted to several private companies. Despite all the challenges we dealt with, it is a pleasure to present such a robust study," explains Ana María Aransay, project coordinator and head of the Genetic Analysis Platform of CIC bioGUNE.

➤ **Combined information of thousands of genetic variants**

A Single Nucleotide Polymorphism (SNP) is a variation that occurs when a single nucleotide (A, T, G or C) in a specific position in the genome differs among the members of the same species. It is believed that there are some 10 million SNPs distributed in the 3,000 million nucleotides that make up the human genome. Thus, even individuals from the same population may have different nucleotides in the same position. Although variable, SNPs are relatively stable over the course of evolution, that is, they do not change much from generation to generation, making them valuable genetic markers for population studies. Generally, the differences in allele frequencies are used, i.e. the proportion of a particular nucleotide in each SNP for each population.

In this study, a total of 60,000 SNPs spread throughout the genomes of 240 Europeans were analysed, of which 83 are Basques from Spanish regions and 24 are Basque from French regions. For each population, allele frequencies were measured for each of the SNPs and compared. As an example, for a given SNP we can observe that nucleotide A is more common in the Basque population (allele frequency 60%), and that in the other population nucleotide C is found with higher frequency (allele frequency 80%).

These calculations are performed for each SNP using bioinformatics tools capable of analysing millions of data simultaneously.

It should be noted that differences in allele frequencies of a single SNP do not provide any information alone; however, the combined analysis of a large number of SNPs (60,000 in this case) is highly effective when it comes to determining the genetic differentiation of populations.

➤ **Population differentiation**

The study concludes that Basque population differs genetically from the rest of Europeans, as much as, for example, the Italians from the Russians. Although individuals from other parts of Spain have not been analysed, the results suggest that the Basque population is as different from them as from other European populations. The differentiation of the Basques with respect to, for example, French people, despite their geographic proximity, shows that the Basques have historically been more isolated than most European populations – similar to what could have happened with, among others, the Sardinians.

"The study suggests that there is no special 'particular' genetic feature in the Basque population compared with other regions, as the Basques can be as 'particular' genetically speaking as the Sardinians, Orcadians, Russians, etc.", states Naiara Rodríguez-Ezpeleta, researcher of CIC bioGUNE who participated in the study.

"The conclusion of the study is that the Basques are as different from other European populations as those European populations that are considered to be isolated, i.e., that Europe is divided into groups and one of these groups are the Basques", says Rodríguez-Ezpeleta.

These findings clearly differ from another study recently published in the same scientific journal (*Human Genetics*), carried out by a research group at the Universitat Pompeu Fabra, Catalonia (Laayouni et al. Hum Genet 2010).

According to that study, the Basques from Spanish regions are more like individuals from other Spanish regions than the Basque-French, and the Basque-French are more like other Europeans than Basques from Spanish regions. The research also highlights that Basque-Spanish people do not differ notably from other Spanish regions, i.e. the genetic distance between the Basques and citizens from other regions is no greater than the distance observed between other pairs of regions.

The study carried out by the Basque research consortium, however, overturns these conclusions and confirms scientifically that the Basque population (Spanish and French) is a genetically homogeneous group clearly distinguishable from other European populations.

Researchers who led this new study were surprised when they found out the conclusions of their colleagues in Catalonia, because, in their opinion, these conclusions do not tally with scientific evidence recognised by the scientific community itself. They believe that the results of the Catalan researchers are due to the fact that in their analysis genetic data from an average of a group of individuals were used and not that of each individual, which paints a less reliable picture. They only used 109 genetic markers on an individual basis compared to the 60,000 that have been used in the later study.

"They have conducted their study with more markers than us, but with a group genotype. Out of the 240,000 markers used, only 109 were treated on an individual basis. They used a large number of markers, but genotyping is done on a group level and not individually, which prevents it from having the accuracy of our genotyping study, which was conducted on an individual basis," concludes Rodríguez-Ezpeleta.