

SCIENTIFIC INFORMATION FOR THE WORKSHOP

Genetic research into the ultimate causes of diseases in humans is a key scientific objective for third millennium medicine. With the completion of the first full euchromatin map (Human Genome Project) and the subsequent genetic variation maps developed in phases I and II of the HapMap project, we have obtained enough basic information to launch the first genome-wide scan projects for the most significant diseases in humans.

Although we are currently beginning to understand how genome variations affect the risk of suffering all diseases, the key genes that influence each disease are almost entirely unknown. The answer must lie in understanding the degree of variation existing among individuals, which may be as high as 10% (if we consider the gene dosage) at each locus, and whose relation with healthy and unhealthy states is almost completely unknown.

This means that we are experiencing a moment of biotechnological development in which it is crucial to decode the genetic variation structure of a specific population in order to complete genetic studies that are competitive, permit the development of accurate diagnostic tools for the population studied, and may even be universal enough for use among other populations. In the same way, decoding and correlating this information will permit the design and preparation of far more precise treatment methods (personalised medicines) that will revolutionise the current pharmacological perspective.

In this context of development and innovation, currently known as the *Post-Genomic Era*, NeoCodex presents the first haplotype variation map produced in Spain. This information has been generated using material from our biobank, now one of the most extensive and detailed DNA banks in Spain. NeoCodex contains 45,000 specimens from 15,000 individuals born in Spain. Each of these individuals has at least two previous generations (on the paternal and on the maternal side) registered in this country. From this group, we have selected 1,200 individuals from the general Spanish population with the aim of performing a total of 262,264 genetic tests in this study, using DNA microchip technology and cooperating with more than 20 public and private hospitals all over Spain. More

than 100 researchers have participated in this study in which 314.7 million genetic tests were completed and one of the most detailed haplotype maps in Europe was created. The accuracy of allele frequency and the data on unbalanced binding among the markers obtained (estimated over 2,400 chromosomes) is greater to those registered in the International HapMap Project.

Our project has been co-financed by National Development Agents, such as the Ministry for Education and Science, the Regional Government through the Regional Ministry of Innovation, Science and Business, and the IDEA Agency, as well as by the Andalusian Technological Corporation (CTA). The genomic information obtained, unique in our setting, provides a quantum leap forward in our understanding of the genomic structure of Spanish population, and permits immediate application of these results to research into common illnesses such as ailments linked to individual Cardiovascular Risks, Osteoporosis, cerebral neurodegenerative diseases (Alzheimer's, Parkinson's) and different types of Cancer.

Apart from NeoCodex's objective to make scientific and technical use of the results, which is currently underway, NeoCodex's intent as a biotechnology company is to sign an agreement with the Regional Ministry of Innovation of the Government of Andalusia to disseminate and enlarge this project. The basic aims are to permit free access to this information for all researchers who demonstrate their scientific goals and technical capabilities, as well as to continue expanding the map resolution so as to make this repository one of the largest complete genome databases in the world.

IMPACT ON INNOVATION

Progressing correctly in the post-genomic era requires the collection of general variation data for all populations. Although some pilot programmes exist in Europe, as well as countless unincorporated results, most countries do not have an accurate, organised genomic variation database for their population. In fact, in the case of the most developed countries in this field, such as Germany, France, the United Kingdom and Iceland, waves of immigration affected the haplotype structure of the resident population and the precision of the variation estimates may be lessened.

Having accurate information about the genetic variation of a population such as that of Spain permits more detailed studies on the genetic association of the most prevalent diseases in the country in question to be carried out, thus lowering the development costs for SNP studies (since a large number of controls are already genotyped), and speeding up research into countless genetic biomarkers that are being studied both within and outside Spanish borders. For this reason, having a tool of this type at our disposal will greatly accelerate R&D in this sector, encouraging biomedical research, permitting the development of personalised medicines and pharmacogenetics and placing Andalusia at the forefront of Human Genome research.