



**ΟΜΙΛΗΤΗΣ:** **Dr. Ana Aransay**

Functional Genomics Unit

CIC bioGUNE

Parque Tecnológico de Bizkaia, SPAIN

**ΘΕΜΑ:** **How can we analyse the genome? High throughput technologies**

**ΤΟΠΟΣ:** Αίθουσα Σεμιναρίων ITE/ΕΙΧΗΜΥΘ

**ΗΜΕΡΟΜΗΝΙΑ:** **Τετάρτη, 22 Ιουλίου 2009**

**ΩΡΑ:** **12:00**

### **ΠΕΡΙΛΗΨΗ:**

Since the publication of the first draft of the Human Genome in 2001, there has been a vertiginous development of high throughput technologies for the detailed characterization and the understanding of the genomes performance. The general aim of this revolution is to elucidate the insights of the diversity in the response to certain drug-treatments or the susceptibility/resistance to suffer a particular disease.

Among the designed techniques, there are several ones for high throughput genotyping of single nucleotide polymorphisms (SNPs), which occur every 100-300 base pairs throughout the approximately 3,300 million that the human genome contains and, therefore, they are responsible for about 90% of our genetic variation. Several companies have standardized arrays which allow the characterization from 96 to more than 2 million SNPs along the whole human genome. Some of these designs have also included probes (oligonucleotides) to detect deletions and/or duplications at chromosomal level (copy number variations, CNVs), increasing the cost-efficiency of these methodologies.

For the fully comprehension of the human genome, the knowledge of mRNA and smallRNA expression levels in affected tissues is critical. Therefore, several companies offer the possibility to study mRNA and/or smallRNA contents at whole genome or regional level. In addition, epigenomic modifications, such as methylation, are proved to be regulating certain genomic regions (i.e. genomic imprinting). Therefore, methodologies for whole genome analysis of methylated regions in the targeted tissues are available.



During the last 5 years, another technological revolution is helping to explain complex cellular behaviors based on the deep sequencing of big genomic regions (i.e. 3 Gpb), whole genomes and transcriptomes, as well as the complete batch of DNA interacting with particular proteins.

All these techniques generate huge amounts of data that should be properly stored. Therefore, computing workstations, servers and data bases should be taken into account in the labs running this sort of projects. Once raw results are saved, the first step of the data analysis is to define quality control thresholds and inclusion criteria and filter out those sets (samples and/or markers) that do not accomplish the established conditions (i.e. minor allele frequency and Hardy-Weinberg equilibrium in genotyping strategies). Once good quality samples and markers are selected, each project would follow different strategies for the analysis and, thus, correct input formats for the bioinformatic tools in use should be generated.

## **Short Biographical Sketch**

Ana M. Aransay received her B.Sc. degree in Biology (speciality: Fundamental Biology) from the University of the Basque Country (UPV/EHU) in 1992. She obtained her Ph.D. at the Department of Clinical Bacteriology, Parasitology, Zoonoses and Geographical Medicine at the Faculty of Medicine of the University of Crete (Greece), under the supervision of Prof. Yannis Tselentis. During the Ph.D. (grant from the Basque Government) she became familiarized with Molecular Biology techniques (isoenzymes, cloning, sequencing, PCR, PCR-RFLP, etc.) and also with diagnosis, typing, phylogeny and epidemiological studies.

After receiving her PhD in 1999 she obtained two consecutive post-doctoral research grants from the Basque Country Government to carry out a project on population genetics of vectors of parasitic diseases. The post-doctoral research work was performed from 1999 to 2004, first at Entomology Department of the Natural history Museum of London (U.K.), under the supervision of Dr. Paul D. Ready and later at the National Centre of Microbiology of the Instituto de Salud Carlos III in Madrid (Spain), under the supervision of Dr. Ricardo Molina.

Today she is in charge of the Genome Analysis Platform at the Functional Genomics Department of CIC bioGUNE ([www.cicbiogune.es](http://www.cicbiogune.es)).