

## Our Company

Genomnia is a start-up SME established in October 2008 with the aim of offering deep sequencing and bioinformatic analysis services. Recently Genomnia progressed from Solid to Ion Torrent sequencing technology. We offer a wide array of services to private and public institutions, which include genome and exome analyses, personalized gene panels, transcriptome analysis, genome-wide epigenetic analyses (ChIP-seq and Methyl-Seq), and metagenomic analyses. In addition to the expertise in molecular biology, we have relevant experience in bioinformatic analysis of deep sequencing data applied to biomedical or molecular genetic projects.

The 300 m<sup>2</sup> of Genomnia laboratories are equipped with updated instruments and our experienced personnel will take care of the planning, optimization and analysis of your sequencing projects.

Genomnia technological platform is composed of the Life Technologies Ion Personal Genome Machine<sup>®</sup> (PGM<sup>™</sup>) and the ION S5 sequencers. By eliminating the optical system, typical of classical sequencing machines, and exploiting the biochemical reaction caused by the incorporation of a nucleotide into a strand of DNA by the DNA polymerase, ION TORRENT technology provides sequencing that is simpler, faster and highly cost effective. The throughput is up to 15 Gb for 200 bp sequencing or 8 Gb for 400 bp sequencing per run per day. Major applications of this technology are: the detection of mutations and genetic alterations by targeted resequencing (single genes or genomic disease-related panels); human exome sequencing; *de novo* sequencing or viral and bacterial genome sequencing; transcriptome (RNA-seq, miRNA-seq, etc) and epigenetic studies (ChIP-seq, MBD-seq, etc.). We also perform human and non-human metagenomic studies with the analysis of the 16S RNA hypervariable regions or the whole genome microbial analysis.

The Genomnia laboratories are organized following the Life Technologies guidelines for the ION TORRENT deep sequencing facility in order to avoid cross-contamination through the workflow:

### **Libraries preparation laboratory**

Nucleic acids extraction, samples quality control and library preparation are performed in this laboratory.

### **Ancillary laboratory: manual emulsion preparation (e-PCR), Digital PCR and Real Time PCR**

A part of the emulsion-PCR process for templated beads (to be successively loaded on the sequencer) preparation is performed in this laboratory. Beads can be prepared by a semi-automated system, composed of One Touch 2 and One Touch ES instruments, or by a completely automated procedure, performed by the Ion Chef instrument.

Real time or digital PCR experiments for relative or absolute expression analyses and the detection of rare allelic variants are performed in this room.

### **Deep sequencing laboratory**

This is the laboratory, there sequencers are located.

### **Bioinformatics Unit**

The aim of the Bioinformatics Analysis Unit at Genomnia is in a first instance the delivery to the scientific partners of high quality ION PGM<sup>™</sup> and S5<sup>™</sup> sequence data, unambiguously mapped to the reference genome, counted with precision, annotated and classified with the maximum level of precision and information content. Current bioinformatic analysis skills include transcriptome analysis, both coding and non-coding, of human and model organisms, with focus on tissue or circulating microRNAs; analysis of exome variants and insertions / deletions for the identification of putative disease genes, including "trio" design; the identification and differential analysis of genome regions with regulatory functions through the interpretation of ChIP-Seq or Methylation experiments; assembly and primary annotation of small genomes; analysis of quantitative PCR. For a detailed description see the descriptions specific for every ION PGM<sup>™</sup> and S5<sup>™</sup> application.