

Our Company

Genomnia is a start-up SME established in October 2008 with the aim to offer deep sequencing and bioinformatic analysis services. Thanks to the experience of the highly specialized personnel, able to give a significant contribution to the planning, optimization and analysis of your sequencing projects, Genomnia can offer to private and public institutions a wide array of services, including not only genome, exome, customized gene panels and transcriptome analyses, but also genome-wide epigenetic analyses (ChIP-seq and Methyl-Seq) and metagenomic studies. Indeed, our company expertise spans from molecular biology to bioinformatics, ensuring the processing and interpretation of deep sequencing data applied to biomedical or molecular genetic projects.

Genomnia workflow takes advantage of Life Technologies Ion Torrent sequencing platform, comprising Personal Genome Machine® (PGM™) and Ion S5 sequencers. These systems exploit the ability to measure even small changes in pH value and apply semiconductor properties to directly detect the DNA-polymerase-mediated incorporation of nucleotides into the DNA growing chain, since this biochemical reaction produces the release of a proton. This allows a direct translation of a chemical information (nucleotide identity, i.e. A, T, C, G) in a simple digital input (0 or 1), getting rid of the optical components required by traditional sequencing for the detection of fluorochrome-conjugated nucleotides: sequencing is thus simpler, faster and highly cost-effective.

Ion Torrent platform ensures a throughput of up to 15 Gb for 200 bp sequencing or 8 Gb for 400 bp sequencing per run per day. Major applications of this technology comprise detection of mutations and genetic alterations by targeted resequencing (of single genes or of genomic disease-related panels), human exome sequencing, *de novo* sequencing of viral and bacterial genome, transcriptome (RNA-seq, miRNA-seq, etc) and epigenetic studies (ChIP-seq, MBD-seq, etc.). We can also perform human and non-human microbiome studies, by either 16S rDNA hypervariable regions analysis or whole meta-genome sequencing.

Our 300 m²-wide laboratories are equipped with state-of-the-art instruments and are manned with experienced personnel. Lab spaces are organized according to Life Technologies guidelines for Ion Torrent deep sequencing facility, to avoid cross-contamination through the workflow and to ensure optimal performances.

Samples and libraries preparation laboratory

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

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

In this laboratory we perform some steps of the emulsion-PCR process to prepare templated beads, that are successively loaded onto the sequencing chip. Beads can be prepared either by a semi-automated process, using One Touch 2 and One Touch ES instruments, or by a completely automated procedure performed by Ion Chef system.

This laboratory is also equipped to perform Real time or digital PCR experiments for relative or absolute expression analyses and for the detection of rare allelic variants.

Deep sequencing laboratory

This is the laboratory where Personal Genome Machine® (PGM™) and Ion S5 sequencers are located and where sequencing is performed.

Bioinformatics Unit

The aim of Genomnia Bioinformatics Unit is providing customers and scientific partners with high quality Ion PGM™ and S5™ sequence data, unambiguously mapped to the reference genome, counted, annotated and classified with the maximum level of precision and information content. Our bioinformatics expertise enables us to perform transcriptome analyses, both on coding and non-coding RNA, of human and model organisms, with a focus on tissue or circulating microRNAs; analysis of variants and insertions/deletions for the identification of putative disease-causing genes within gene panels or whole exome, also arranged as “trio” experimental design; identification and differential analysis of regulatory genomic regions through the interpretation of ChIP-Seq or Methyl-seq assay results; assembly and primary annotation of small genomes; metagenomic studies; analysis of quantitative PCR data. For more information, please refer to the specific ION PGM™ and S5™ service module.