**Bioinformatics steps for personalized genome analysis**

1. **Genome analysis:** DNA and RNA sequences from different experimental sources and using various technologies and platforms
2. **Consequences of mutations and genomic alterations:** Mutations in coding and non-coding regions, changes in gene expression, genome structure alterations
3. **Network level analysis:** Metabolic and signaling pathways, gene control networks and functional classes
4. **Drug related to proteins and pathways**
5. **Collaborative interfaces**

**Principal developments**
- Software for management and analysis of NGS and other high-throughput genomic data
- Methods for the prediction of consequences of mutations at protein family level
- Methods for the prediction of binding sites for transcription factor and miRNAs
- Analysis of alterations in splice sites and splice factors
- Prediction of the consequences of alterations in epigenetic markers
- Databases and analysis platforms for pathways and networks
- Systems linking disease and symptoms to molecular entities
- Emerging simulations of biological networks and pathways
- Public repositories of drugs and small molecules
- Systems linking protein networks with drug targets
- Initial systems able to display genomic information and complex analysis in biomedical/clinical environments

**Key problems**
- Data storage and organization
- Linking heterogeneous data types
- Adapting algorithms, methods and tools to the fast evolution of the experimental techniques
- Definition of function at various levels, from biochemical to cellular
- Collection of accurate experimental data on consequences of mutations
- Prediction of activating and deactivating mutations at a quantitative level
- Integration of pathway information
- Completing pathway and network data with experimental information
- Standards for pathway simulation and validation
- Accessibility of medical information and toxicology reports
- Adaptation of text and database mining system
- Extraction of accurate information on in vivo drug targets
- Data protection
- Data provenance and traceability
- Robustness and performance
- Adequate information for users and different level of access

**Emerging simulations of biological networks and pathways**

**Public repositories of drugs and small molecules**

**Systems linking protein networks with drug targets**

**Collaborative interfaces**

**Initial systems able to display genomic information and complex analysis in biomedical/clinical environments**