

## Future Challenges of Biomedical Informatics for Translational Medicine

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### Abstract and Objective

There are many challenges for Biomedical Informatics (BMI) to meet the promises of Translational Medicine. Several of the challenges have to do with the re-use of clinical data to progress towards 4P Medicine (personalized, preventive, participatory and predictive) and using epidemiology and clinical research as well as the translation of data and knowledge obtained in the laboratory into useful information for the clinical setting. The INBIOMEDvision EU funded project tackled these challenges in a series of events with expert scientists in different related disciplines. As a result, some consensus ideas were reached about the BMI challenges in the research areas of Genotype-Phenotype integration, Translational Systems Biology and Re-use of Clinical information in the Electronic Health Records (EHR) for research and what is needed to meet them.

### Keywords:

Biomedical informatics, translational medicine, personalized medicine

### Methods

Organisation of activities in the framework of INBIOMEDvision, a Coordination and Support action funded by the European Commission under the Seventh Framework Programme (FP7/2007-2013) and COMBIOMED (MINECO). They comprise Think Tanks focused on “Genotype-Phenotype integration” (Gen-Phen), “Translational Systems Biology and Bioinformatics” (TSBB), “Re-use of Clinical Information for Research” (RCIR) and a two-day meeting “Integrative Knowledge Management for Empowered Healthcare and Research”, where experts from different disciplines discussed and shared their ideas relevant to the BMI challenges to foster further advances in translational medicine.

### Results

Despite the years of work in BMI, the discussions held by the experts in the different activities highlighted some remaining challenges that still need to be met. A number of guidelines were proposed to meet these challenges.

### Challenges for BMI

In Gen-Phen research, the knowledge obtained is large, complex, and with uncertain conclusions, and not readily suitable for packaging and displaying it for clinical application or train-

ing or for its integration in EHRs. These reasons, together with a lack of training and information of physicians about the nature, limitations and potential of modern genotype-phenotype knowledge, are hindering the application of research findings in the clinical setting. The challenges of the research area of TSBB focus on the analysis of data from high throughput studies (GWAS, PheWAS, EWAS) to obtain bioinformatics signatures in molecular diagnostics, prognosis, drug response and discovery, and on establishing reliable and reproducible computational models at different levels from molecules to population. With respect to the RCIR the challenges are to unify and standardize information in the EHR and to determine minimum required data to ensure better quality of data entry or capture, while paying attention to quality assurance. Another important goal is to ensure interoperability of IT across EU as well as to promote unification of regulations across EU regarding EHR (legal, ethical, security and confidentiality issues).

### Trends for BMI Research to meet the highlighted Challenges

Development of standards and of validated repositories and specific tools and resources based on genotype/phenotype associations for patient stratification in clinical trials, are both common research needs. In addition, standardization of methodologies and database formats, and application of crowdsourcing and collaborative-competition in system biology, is crucial. Among the trends identified for EHRs are, the use of standards, the digitalization and standardization of data capture to allow better quality and optimization for data sharing between hospital and community care settings (i.e. EHR4CR) enabling its re-use for research purposes, the increased automation of data entry facilitating both clinicians' and the clinical community's work and the involvement of clinicians of different lines of work. The horizontal challenges of BMI fall into two categories: i) ICT data quality and validation that would include data management and integration, data security and development of user interfaces; and ii) socio-economic challenges, that would include openness and information sharing, security and privacy, training of physicians, patient empowerment, industry-academy collaboration, and intellectual property management.

### Conclusion

There are still some key areas of BMI to research and promote to further Translational Medicine in the future.