

## Translational informatics

Development of a computational infrastructure dedicated to the integrated analysis of data from "-omics" sciences (cf. [www.omics-ethics.org/fr/definition-science-omics](http://www.omics-ethics.org/fr/definition-science-omics)) related to human genetic diseases and health. This infrastructure includes algorithms and original methods, such as:

- Algorithms for integrative structural biology, in the context of ongoing funded projects, including the European INSTRUCT research infrastructure for 'Integrative Structural Biology' and the French ANR Investments for the Future project: FRISBI, coordinated by Bruno Klaholtz, IGBMC, Strasbourg.
- Multi-scale methods for a better understanding of protein properties (structure, interactions, dynamics...) and better integration of the heterogeneous data associated with a protein or a family of proteins. Ongoing project, BIPBIP, funded by French ANR in collaboration with Annick Dejaegere, IGBMC, Strasbourg.
- Evolutionary analyses of protein sequences from NGS data, applied to health, in collaboration with the [Data Mining](#) data mining and [SONIC](#) teams.
- GREMSAP (GRid Evolutionary Multiple Sequence Alignment Platform), in collaboration with the [SONIC](#) team, l'Institut des Systèmes Complexes Paris Île de France, l'Institut de Neurobiologie Albert Fressard.
- Social Network Clinical Database for Intellectual Disabilities: Development of a social network for patients, related to genetic diseases causing developmental disabilities, in collaboration with the [SONIC](#) team, the [Translational medicine & neurogenetics team, IGBMC](#), led by Jean-Louis Mandel, and the [<http://www.ucad.sn> Cheikh Anta Diop University, Senegal].
- Infrastructure for 'big data' management for the translational analysis of mutations involved in human genetic disease. BIRD/SM2PH-central is aimed at the integration of heterogeneous data (genomic, phenotypic, evolution, cellular networks,...), with data mining methods (association rules, inductive logic programming,...), and includes the design of a semantic query language (BIRD-QL) and the development of original graphical interfaces. These developments are done in collaboration with Hoan Ngyugen, IGBMC, Strasbourg.

## Systems bioinformatics

Development of research in the field of biological systems analysis, to understand genotype-phenotype relationships, notable concerning genetic diseases, and the study of complex biological systems, for example in various cancers or rare diseases (ciliopathies, myopathies,...) in close collaboration with the Human Genetics Laboratory, led by H el ene Dollfus ([www.unistra.fr/index.php?id=19264&L=3](http://www.unistra.fr/index.php?id=19264&L=3)). The originality of our approach lies in the extraction and exploitation of evolutionary information (sequence analysis, comparative

## On-going projects

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genomics, etc.) in order to improve the analysis of the different levels of complexity in biological systems. Some examples of systems-level projects:

- Analysis of macromolecular complexes (TFIID...) in collaboration with the [Department of Integrative structural biology, IGBMC](#).
- Analysis of organelles (exosome, endosome...) in collaboration with the [membrane traffic and lipid signaling team, GMGM](#): synthetic yeast evolution, the characterization of the shared / specific features at different levels (nucleotide, gene, pathway), the identification of correlations between the evolutionary scenarios and the modifications identified in the molecular processes.
- Genomic analysis of 1000 myopathy patients (Myocapture project and ongoing FRM-funded project, in collaboration with the team of Jocelyn Laporte, IGBMC) with the goal of identifying and characterizing new genes in these diseases.
  - Analysis of genomic data from ciliopathy patients (Bardet-Biedl Syndrome and Alström Syndrome) to identify the genes responsible for the diseases, and to understand the links between genotypes/phenotypes, in collaboration with [Human Genetics Laboratory](#), led by Hélène Dollfus.
- Integration of "omics" data to develop a new generation of the Vaccinia virus (VACV). Ongoing funded project (OncoVaccine: ANR Recherches Partenariales et Innovation Biomédicale) in collaboration with TRANSGENE, the HTCS platform led by Laurent Brino, IGBMC and the team of Etienne Weiss, IREBS, Strasbourg.
  - ImAnno is a collaborative project between the LBGI and teams at the IGBMC, Strasbourg (P. Dollé) and the Institute of Vision, Paris (J. Sahel, T. Leveillard) aimed at developing an ergonomic and integrative annotation tool for biological images (ISH, fundus...). Once annotated, the images allow to access all the knowledge extraction tools implemented or developed by the LBGI, including protein-protein interaction networks, tools for automatic analysis of the evolution, transcriptomic data, etc.
  - Multi-scale tempo-spatial modeling of the primary cilia as a means of communication (intracellular transport, physical cell-cell interactions, environmental sensory system) and its role in the cell cycle, development, as well as the evolution of eukaryotic organisms, in collaboration with the [Digital Campus for Complex Systems](#).