

Structuring Knowledge on Human Diseases: Challenges and Opportunities

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Abstract

Recent technological breakthroughs are producing an unprecedented volume of data on the genetic determinants of human diseases. In our quest to identify disease biomarkers and to support drug discovery projects, it is necessary to place this wealth of information in the context of current biomedical knowledge. In this talk I will present our work in this area exemplified by the DisGeNET¹ and PsyGeNET² knowledge platforms. The key role of text mining for extraction of relations between biomedical concepts will be discussed³. I will present some of the hurdles we face in structuring the knowledge of human diseases, such as the prevalence of data silos in biomedicine, limitations of current ontologies and the need of data curation and prioritization strategies. In this regard I will share our experience for data curation leveraging on users' community and crowdsourcing. Finally, the importance of building an ecosystem of linked data in biomedicine to support translational research will be presented.

Biography

Laura I. Furlong is head of the Integrative Biomedical Informatics Group, which belongs to the Research Programme on Biomedical Informatics (IMIM-UPF) and Associate Professor at the University Pompeu Fabra at Barcelona (Spain). She has a PhD in Biology from the University of Buenos Aires and a Msc in Bioinformatics by University Pompeu Fabra. She has a broad expertise covering molecular biology, computational systems biology and text mining. Her current research lines include: a) development of new strategies and tools for knowledge extraction from

biomedical literature; b) network biology for the study of human diseases and drug toxicity; c) bioinformatic approaches for the reuse of clinical data for research. Her group also maintains knowledge resources to support translational research, such as the databases DisGeNET and PsyGeNET. She has published over 40 peer-reviewed articles, and act as reviewer for the journals *Bioinformatics*, *BMC Bioinformatics*, *BMC Systems Biology*, *Database* and *PLOS* journals. She has participated in several FP7 projects (@neurist, EU-ADR) and is currently involved in the IMI (Innovative Medicines Initiative) projects eTOX, OpenPHACTS, EMIF and IPiE and the H2020 project MedBioinformatics.

¹<http://www.disgenet.org/>

²<http://www.psygenet.org/>

³<http://ibi.imim.es/tools/befree/>