LUXEMBOURG INSTITUTE OF HEALTH
RESEARCH DEDICATED TO LIFE

UNIVERSITÉ DU LUXEMBOURG

LECTURE SERIES & WORKSHOPS

TRANSLATIONAL BIOINFORMATICS AND SYSTEMS BIOMEDICINE

13
FEB. 2019
Wednesday

LECTURE
Maison du Savoir
MSA 2.380
(2, avenue de l'Université
L-4365 Esch / Alzette)
4.00 - 5.00 pm

MEET THE SPEAKER*
Light snacks provided
Maison du Savoir
MSA 2.390
5.00 - 6.00 pm

1 2 3 4 5 6 7 8

*Please register by sending a mail to florence.henry@lih.lu

SPEAKER
Dr Laura I. FURLONG
Head of the Integrative Biomedical Informatics Group
Associate Professor at UPF
Research Programme on Biomedical Informatics (GRIB)
Hospital del Mar Medical Research Institute (IMIM) DCEXS, Pompeu Fabra University (UPF), Spain

HOST:
LIH

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THE DisGeNET KNOWLEDGE MANAGEMENT PLATFORM FOR DISEASE GENOMICS

ABSTRACT

In the last few decades, our knowledge about the genetic underpinnings of human diseases has grown at an unprecedented pace. Advances in sequencing technologies have made it possible to catalog a large volume of genetic alterations found in patients for a broad spectrum of diseases. Data resulting from GWAS studies, experiments in animal models of disease, and from NGS sequencing pipelines are publicly available, but scattered across different repositories. Furthermore, accessing, navigating and analyzing this information is a challenge due to its heterogeneity and lack of standardization. To enable translation of this wealth of knowledge into better disease biomarkers and drug therapies, this data should be made readily available to researchers and clinicians.

The DisGeNET knowledge platform aims to support translational applications by providing a unified catalog of disease associated genes and variants, along with a suite of tools to exploit this information. 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, the need of data prioritization strategies, and the key role of text mining for extraction of knowledge from the literature. Examples of application of DisGeNET data in the area of drug discovery and disease genomics will be presented. Finally, the importance of building an ecosystem of linked data in biomedicine to support translational research will be presented.

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