CISSP/ICPHC 2018 MONTREAL



15th Anniversary of the Completion of the Human Genome Project: Challenging Transition from 'OMICS' Discoveries to Responsible Application

PROPOSED THEMES

* THEME 1: A WIDE RANGE of '-OMICS' DISCIPLINES

15 years after the completion of the human genome project in 2003, have been marked by the application of genomic knowledge to daily life and development of this project has set the stage for rapid development of **`-omics'** laguages - **Transcriptomics, Proteomics, Metabolomics, Epigenomics, Metagenomics, Vaccinomics** and so on and so forth.

- This theme will focus on the application of the following '-omics' languages for personalized medicine:
- <u>Genomics</u> plays an important role in disease processes and with the technological advancements made over recent years there is a growing use of genomic tools to guide clinical decisions.
- This theme will provide an excellent platform for scientists moving from GWAS into biological function and disease mechanisms by bringing together a multidisciplinary group of scientists, bioinformaticians, clinicians as well as technology developers interested in using genomic advances to develop and deliver therapies for patients with common and rare diseases.
- **Epigenomics** has emerged as a new multi-disciplinary discipline focusing on interactions between genomics and environment in disease prediction, prevention and treatment. Though associations between epigenetic variation and a variety of risk factors with the development of many diseases continue to emerge, in many cases, the causality has not yet been established.
- This theme will focus on epigenomic studies across a wide range of human pathologies, such as cancer, cardiovascular diseases, diabetes, aging, and neurodegenerative diseases. It will also explore technological and methodological progress providing a forum to present and discuss recent advances in epigenomics. Moreover, the theme will emphasise recent studies on computational epigenomics and regulatory networks with a special attention on approaches needed to demonstrate causality of epigenomic control of diseases.

- Metagenomics is one of the fastest growing disciplines linking many non-infectious diseases with bacterial populations in the gut. Microbiote could be considered as an environmental factor modulating the host metabolism and includes more than 100 trillion cells of 400 species (approximately ten times the total number of cells in the human body). Alterations and perturbations in microbiome composition appear to be important for metabolic disorders, such as obesity, T2D, cancer as well as inflammatory bowel conditions and autoimmune diseases. Therefore, manipulation of the gut microbiota may be an important therapeutic strategy to regulate energy balance in individuals. However, since every individual is different, gut microbiota should be evaluated at personal level.
- This theme will update our current knowledge of how various microbiomes in gut communicate with the host and the factors that influence these interactions and will discuss the new developments enabled by metagenomics.
- <u>Metabolomics</u> is the global analysis of a large number of metabolites providing a wealth of information about the biochemical status of cells, tissues or organisms. It also plays an important role to elucidate the function of novel genes.
- This theme will focus on metabolomics workflows (methods, technologies and data treatments), the needs, challenges as well as interpretation of metabolite correlations for personalized medicine application.
- **Proteomics:** provides functional information of key regulating protein target(s) in disease development which is a crucial step in personalized medicine.
- This theme focuses on the benefits and potential use of personalized proteomic analysis for the diagnosis and treatment of disease. It will also highlight essential topics of protein expression and analysis, mass spectrometry in proteome research, proteomics database and proteomics from discovery to function for personalized medicine application.
- <u>Transcriptomics</u>: With the emergence of RNA sequencing (RNA-seq) technologies, RNAbased biomolecules hold promise for their diagnostic, prognostic and therapeutic application in various diseases, including cancers and infectious diseases. It is potentially instrumental, jointly with genomics in causality exploration of functional pathways in health and disease and human evolutional variance among geo-ethnic groups.
- > This theme will focus on recent advancements in molecular profiling research and technology, such as RNA-seq as applied to clinic and prediction of future of personalized medicine in complex diseases
- <u>Vaccinomics</u>: The current medical practice in vaccinology is to administer the same set of vaccines to everyone in the population. However, the major weakness of this approach is that ignorance in individual variability for disease risk immunologic response, and any genetic propensity for reactogenicity, as well as differences in dose amount needed to generate immunity. To this end, the field of pharmacogenomics could provide a promising role for vaccine research and development of novel personalized vaccines together with novel vaccine adjuvants as a new public health tool in designing safer and more effective vaccines for high and low income countries.
- > This theme will focus on the role of HLA genes, cytokine genes, and cell surface receptor genes for genetic polymorphism leading to individual and population variations in immune responses to vaccines.

***** THEME 2: BIG DATA – ARTIFICAL INTELLIGENCE for OMICS

- Data are central to '-omics' research and practices, however, due to problems in data definition, formats, structures and semantics, it is difficult to integrate and analyze this exponential growth of data. Thus, efficient management and analysis of this heterogeneous and complex set of data, as well as extraction of valuable information to be translated into clinical practice are extremely important
- Artificial intelligence which has significantly gained grounds in our daily livelihood is transforming the world of medicine. Machine learning is often described as a sub-discipline of AI, that is showing the most promise at providing tools that medicine and industry can use to predict the future and to focus them on solving real-world problems with neural networks designed to mimic our own decision-making processes. Essentially Deep Learning involves feeding a computer system a lot of data, which it can use to make decisions about other data. This data is fed through neural networks, as is the case in machine learning. While the traditional approach to solving problems with computers is to give computers some rules and to apply computing force, with machine learning, only data are given and the rules are developed by the computer. Algorithms derived from machine learning can help health providers make faster and more accurate prediction on the risk of a disease in time to prevent it. And also, it can help researchers to understand how genetic variations lead to disease.
- Being relevant for research scientists, clinicians, bioinformaticians and data users interested in integrating genomic results into the clinical setting, this Theme will focus on recent developments in analytical tools and resources that are facilitating big data integration, exploration, clinical advice utility and ethical issues raised by data sharing and usage for personalized medicine.

* THEME 3: CURRENT AND FUTURE APPLICATIONS OF PERSONALIZED MEDICINE

- Current efforts to understand genomic variability in the clinic are systematically moving genomics from discovery to implementation as an evidenced-based strategy for improving the use of medications, risk prediction, thereby providing an important cornerstone for personalized medicine. To accelerate this progress, further work is needed to characterise new disease mechanisms, drug therapeutic targeting. Also the already available options of genomics should be carefully implemented in clinical practice and should not be postponed until the new paradigm arrives.
- This theme will showcase recent advances in genomics, genome editing, direct genome engineering (e.g. CRISPR) disease mechanisms, risk prediction and pharmacogenenetics for personalized drug therapy. Moreover, the growing use and current challenges of merging electronic health records with genomic data in clinical practice will be discussed.

***** THEME 4: PUBLIC HEALTH GENOMICS

- One should not forget that one of the important strategies of human Genome project is "genomics for society" which is important for the integration of genetic knowledge into our everyday life. However, the much-needed public health genomics policy is still missing in many countries including Low Middle Income Countries (LMICs). Reimbursement for genomic testing and genomic-driven interventions as well as legal/ethical/social aspects, availability and qualification of commercial drug tests on the market must also be considered. Genomics should be viewed through a lens of population and public health to move beyond a simplistic focus on biotechnology. Hence, education of health professionals on genomics as well as regional capacity to design rational genomic studies and interpretation of genomics tests will be crucial to the integration of genomics into mainstream health care for predictive and personalized medicine.
 - ✓ GE³LS (Genomics and its Ethical, Environmental, Economic, Legal and Social aspects) By gathering researchers together come from many disciplines in social sciences and the humanities, GE³LS research investigates questions at the intersection of genomics and society in order to help bridge gaps between genomics researchers and other stakeholders.
- An important part of this theme will be to engage the public in discussions around genomic studies providing a forum of discussion of societal, educational and regulatory issues surrounding personalized medicine application together with patient care. Moreover, the Theme will emphasise the need of involvement of scientists from social fields into research and systems analyses within personalized medicine for which there is still lack of readiness for this challenging interdisciplinary and transdisciplinary approach. Finally, this theme will raise several important questions related to the environment, government policy and public perception, among other broad areas of GE3LS research