### **Genomic Medicine in 2015** Proceedings of 2011 Conference

# **BCCGN Conference 2010 Open Remarks and Keynote Address**

Keynote Address – "Life with my sweet son Becket who happens to have an extra chromosome"

After brief introduction, renowned TV presenter Tamara Taggart shares a mother's view of Down syndrome, including her personal journey with her son Beckett. Tamara will highlight some of the sensitivities to be aware of and will provide helpful pointers and tips for communicating with parents.

# Advances in Human Genetics - What You Need to Know

Dr. Jan M. Friedman explained how new genomic technologies such as whole genome sequencing (WGS) promise to identify the underlying cause of many genetic conditions or pharmacogenetic effects. They are already used in research studies of genetic disease and are being introduced into the clinic. The clinical application of this technology will increase as costs decrease and it becomes more widely available. However, genomic technology comes with challenges, including the vast amount of data generated and our inability to interpret it all. This session reviewed the latest sequencing technologies and discussed their clinical applications and possible limitations.

#### Hope of Hype - Pharmacogenomics to Improve the Safe Use of Drugs

<u>Dr. Bruce Carlton</u> talked about how pharmacogenomics is already impacting clinical practice and how there are many examples of it currently being applied at the bedside, including how it is transforming practical clinical drug dosing strategies. However there are certain barriers that remain to the advancement of pharmacogenomics from the bench to bedside care. This session will discuss the latest advances in pharmacogenomics and their advantages, as well as issues to their uptake.

#### Genomics in the Digital Era- a Tool Towards Treatment

This presentation by Dr.'s Clara van Karnebeek and Sylvia Stockler-Ipsiroglu of the BC Children's Hospital showed how disorders causing developmental delay, intellectual disability and epilepsy (IDE) that affect up to 3% of the general paediatric and adult populations are often undiagnosed. The underlying cause often remains unidentified due to limited availability of biomarkers and time consuming, costly diagnostic tests. This session will inform participants about the treatable genetic disorders that cause IDE, the challenges in identifying them, as well as how they are treated. Participants will also learn how genomics facilitates timely diagnosis and initiation of treatment and how it can be combined with digital applications to improve patient outcomes.

#### Healthcare Unwired - Wireless Health Unlocks the Promise of Personalized Medicine

Shauna Turner MBA, Managing Partner of Strategic Incite presented on how Wireless health has been heralded as the ultimate vector for delivering on the promise of Personalized Medicine. As advances in genomics continue, the convergence of health and digital technologies will lead the way for changes in patient diagnosis, treatment and health management, and ultimately, better outcomes. This session will focus on anticipated changes and impacts on medical practices as a result of technology adoption in genomics and health information technologies and will profile current areas of research and program development.

**Debate Resolution** - Physicians Should Determine the Nature and Extent of Whole Genome Testing Results Conveyed to Patients

**For: Dr. Sylvie Langlois** clinical geneticist at the Children's and Women's Health Center since 1989 and is a Professor in the Dept of Medical Genetics at UBC.

**Against Dr. Mike Burgess,** Professor and Chair in Biomedical Ethics at the W. Maurice Young Centre for Applied Ethics and the Department of Medical Genetics at the University of British Columbia

This debate provided an overview of how the use of new genomic technologies is likely to uncover many unexpected genetic changes that have important medical or social implications unrelated to the disease for which the testing was obtained. The debate discussed what should be done with unexpected findings and who should decide how much is disclosed to the patient or family.

#### Genetic and Genomic Testing - for Cancer Diagnosis and Therapy

Dr. Aly Karsan, MD, Professor of Pathology and Laboratory Medicine, UBC, Medical Director of the Cancer Genetics Laboratory, and Head of Clinical Diagnostic Genomics at the Genome Sciences Centre, BCCA showed how genomic technology is currently used in a wide array of diagnostic tests for recognition of disease. The area where the genomics technology is most used currently in clinical practice is in cancer. This session will cover the role genetic testing plays in cancer diagnostics, cancer therapy, as well as the future of genomic testing in cancer.

#### Personalized Medicine - Value for Money?

Dr. Carlo Marra, Associate Professor in the Faculty of Pharmaceutical Sciences at UBC and a Tier II Canada Research Chair in Pharmaceutical Outcomes talked about how personalized medicine promises to increase the quality of clinical care and reduce health-care costs. Yet only a handful of diagnostic tests have made it to market. The biggest hurdles are not scientific, but relate to economics and the drivers and incentives for change in healthcare providers. Participants in this session will learn about current trends in health care spending in Canada, value for money in the context of health care and the potential impact of genomics on health care costs and outcomes.

# Genomic Medicine and the Clinic - Ethical Implications

Dr. Barbara McGillivray MD, who is a professor in the Department of Medical Genetics at UBC and a clinical geneticist in the Department of Medical Genetics, Children's and Women's Health Centre of British Columbia helped throw light on the ethical implications of clinical genomics. The increasing desire to use genomic technologies for clinical purposes raises many ethical issues including information privacy, the rights of family members and the possibility of discrimination. Session participants will learn to identify ethical issues associated with genomic medicine and outline risks and benefits associated with its use in the clinic.