Pathways to the Clinical Implementation of 
Genome-Wide Sequencing for Rare Diseases: 
Community Consultation on Genome Canada’s Precision Health Strategy

Sunday, June 23, 2019
4:30 PM – 5:30 PM Eastern Time
Strategy Room 1
Sheraton Hotel

Dear Colleagues,

Genome Canada will host a panel discussion on the impact, challenges and opportunities for the implementation of genome-wide sequencing (GWS) as standard of care for rare diseases. Genome Canada has launched a national precision health strategy focused on the clinical implementation of genome-wide testing for rare diseases. Provinces are in advanced planning to repatriate exome and panel testing from US vendors in order to drive local adoption of diagnostic sequencing. With repatriation there are substantial clinical, regulatory and infrastructure challenges to rolling out a high-quality service that meets the needs of patients, health-care providers and other stakeholders.

The panel will introduce Genome Canada’s Precision Health Strategy and address common national themes including:

(1) the disruptive impact of GWS to patients and the Canadian health care systems;

(2) how we should prepare for the transition to whole genomes as routine diagnostic approach;

(3) strategies to empower our gen-omic counselors and clinicians to deal with an order of magnitude more of everything; and

(4) the incredible opportunity for pan-Canadian data sharing to improve patient care through a learning health system.

Finally, the group will address the key question regarding how we as a community can solve these problems in a way that has the most impact for patients in all of Canada, given the fast-pace adoption of GWS.
### Moderator

**Peter Goodhand**  
CEO, The Global Alliance for Genomics and Health (GA4GH)

Peter Goodhand is the Chief Executive Officer of the Global Alliance for Genomics and Health (GA4GH), as well as a leader in the global health sector as a senior executive and board member. Additionally, he has fifteen years of experience as a patient advocate, caregiver, and navigator throughout his family’s battle with a rare cancer.

Goodhand is currently a member of the Occupational Cancer Research Centre Steering Committee, Co-Chair of the Medical and Scientific Advisory Board of Global Genes, Co-Chair of the International 100K+ Cohorts Consortium (IHCC), and a member of the Global Genomic Medicine Collaboration (G2MC) Steering Committee.

### Panel Members

**Cindy Bell**  
Executive Vice President, Corporate Development, Genome Canada

Cindy Bell is Genome Canada’s Executive Vice-President, Corporate Development. She provides leadership in the development and implementation of strategic initiatives and approaches to enhance Genome Canada’s business model and secure funding to support genomics research in Canada.

Dr. Bell joined Genome Canada in August 2000. From 2000-2008 she held the position of Vice-President, Genomics Programs, in which she was responsible for providing policy and strategic advice on scientific and other aspects of Genome Canada’s programs. This included overseeing and managing the peer review process used to establish the research program of Genome Canada.

Prior to joining Genome Canada, Dr. Bell was a Deputy Director in Programs Branch at the Canadian Institutes of Health Research (CIHR) from 1994 to 2000. At CIHR she managed a number of research programs and was involved in policy development and implementation. From 1986 to 1994, Dr. Bell was a researcher at the University of California, Riverside. Her research focused on investigating the basic defect in the genetic disease, Cystic Fibrosis. She obtained her PhD in Genetics from McGill University in 1986.
William T. Gibson  
Professor of Medical Genetics, University of British Columbia  
Senior Clinician Scientist, Laboratory for Obesity Genetics and Indirect Calorimetry (LOGIC), BC Children's Hospital Research Institute  
Investigator, BC Children's Hospital

Bill Gibson trained at the University of Toronto, UWO (now Western University), University of Calgary and at the University of Cambridge. In late 2011 he led the team that was first to publish the identification of rare mutations in the PRC2 complex as causative of Weaver syndrome. Since then, his group has identified additional rare mutations in other epigenetic regulators as causative of both overgrowth and undergrowth syndromes.

He is currently a full Professor at UBC and a Senior Clinician-Scientist at BC Children's Hospital Research Institute. He has also written fictional podcast episodes for Spectral Theatre's Late-Night Double Feature ("Demons of the Helix"), and Darkside Drive ("The Super").

Eriskay Liston  
Genetic Counsellor, The Hospital for Sick Children

Eriskay Liston has been with the Division of Clinical and Metabolic Genetics at the Hospital for Sick Children since 2011. She received her Masters of Science in Human Genetics (Genetic Counseling) from Sarah Lawrence College in 2003, and then worked in the prenatal and cancer genetic programs at New York Presbyterian Hospital for 8 years. Eriskay joined SickKids initially as a laboratory coordinator in the Genome Diagnostic Clinical Laboratory and then joined the Ted Rogers Centre for Heart Research in 2015 as the Genetic Counsellor and Coordinator for the Cardiac Genome Clinic. Eriskay is also actively involved in the University of Toronto M.Sc. Program in Genetic Counselling as a Lecturer, Course Coordinator and Clinical Supervisor.
Christian R. Marshall
Clinical Laboratory Director
Genome Diagnostics, Department of Paediatric Laboratory Medicine
The Hospital for Sick Children

Dr. Marshall completed his undergraduate and graduate training at Simon Fraser University in Vancouver. Following completion of his PhD in 2005, he undertook a postdoctoral fellowship in genetics and genome biology at The Hospital for Sick Children (SickKids). This work focused on copy number variation analysis for discovery of genes involved in risk for developing Autism. From 2009-13, Dr. Marshall was a research associate in genetics and genome biology at SickKids, where he evaluated new genomic technologies and applied them in disease gene discovery across neurodevelopmental disorders. Since 2013, he has held the position of Laboratory Director in the Division of Genome Diagnostics in the Department of Paediatric Laboratory Medicine (DPLM) at SickKids. He has held the rank of Assistant Professor in the Department of Laboratory Medicine & Pathobiology at the University of Toronto in 2015. Between 2014-2018, he completed advanced specialty training in clinical molecular genetics, and was certified as a Diplomat, American Board of Medical Genetics and Genomics (2017), and as a Fellow of the Canadian College of Medical Geneticists (2018) and the American College of Medical Genetics (2018).

Dr. Marshall’s current research focuses on the application of new sequencing technologies for identification of human genome variation and its relation to disease. As a principal investigator and co-director of the Centre for Genetic Medicine, he is exploring the diagnostic utility of whole genome sequencing (WGS) in pediatric medicine and the translation of the technology into clinical diagnostics.

Wyeth Wasserman
Executive Director, BC Children’s Hospital Research Institute
Associate Dean for Research, Faculty of Medicine, University of British Columbia

Wyeth Wasserman, PhD, has worked in the biotechnology and pharmaceutical industry in both North America and Europe, focused on research projects with applied biomedical impacts. His computational biology laboratory develops computer algorithms for the analysis of human DNA sequences.

Originally established at Sweden’s Karolinska Institute and since 2002 at the University of British Columbia, Dr. Wasserman’s lab provides international leadership in the identification and engineered design of DNA sequences that control when and where in the body each gene is active – the On and Off switches. With the arrival of low cost DNA sequencing for patients, his team works closely with clinicians at BC Children’s Hospital to discover the genetic causes impacting patients and families. He has published over 120 peer-reviewed papers, participated in over $15 million of funded research, and supervised more than 60 graduate and post-doctoral trainees.
Dr. Wasserman has served as Principal Investigator or Co-Principal Investigator on nine projects supported by Genome Canada and/or Genome British Columbia. Since 2013, he has served as the Executive Director of the BC Children’s Hospital Research Institute.

Durhane Wong-Rieger
President and CEO, Canadian Organization for Rare Disorders (CORD)

Durhane Wong-Rieger, PhD, is President & CEO of the Canadian Organization for Rare Disorders, Chair of the Consumer Advocare Network, President & CEO of the Institute for Optimizing Health Outcomes, Chair of Canadian Heart Patient Alliance and member of Genome Canada Steering Committee for the Rare Disease Precision Health Initiative. Internationally, she serves as Chair of Rare Disease International, Board member of Asia Pacific Rare Disease International, member of the Editorial Board of The Patient - Patient Centred Outcomes Research, member of the Global Commission to End the Diagnosis Odyssey for Rare Diseases and member of Health Technology Assessment International Patient/Citizen Involvement Interest Group. She is also a certified Health Coach.

Durhane has a PhD in psychology from McGill University and was professor at the University of Windsor, Canada. She is a trainer and frequent lecturer and author of three books and many articles.